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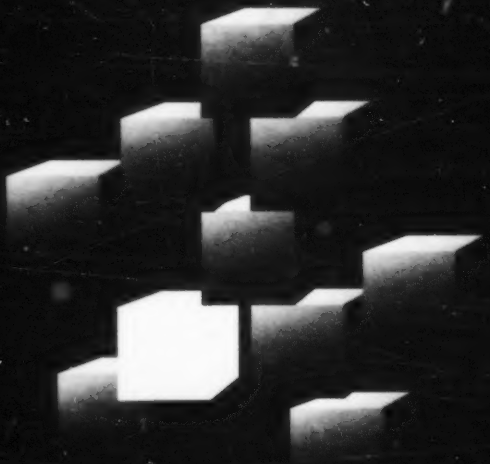
# THE CONSTRUCTION OF DISABILITY AND RISK IN GENETIC COUNSELLING DISCOURSE



THE ROEHER INSTITUTE

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# The Construction of Disability and Risk in Genetic Counselling Discourse

The Roeher Institute

January 2002

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# 1

# EXECUTIVE SUMMARY AND SUMMARY OF RESEARCH

This research initiative set out to assess the ways that the conceptions of disability and risk that circulate in prenatal genetic counselling discourse, law, and public policy affect women's well-being, their decision-making process during pregnancy, and public awareness about people with disabilities. The final goal of this research project is the development of a set of guidelines for genetic counselling that would contribute to a valuation of people with disabilities.

## 1.1 Underlying Concerns

Several concerns about prenatal genetic counselling were at the core of this project:

1) Is there a potential for harm to people with disabilities and to women arising from the predominantly negative attitude toward people with disabilities embedded in some of the assumptions and values that can be implied in or attached to genetic technologies?

2) Is a biomedical conception of health that construes disability as a "flaw" or "defect" to be eliminated or corrected, and that obscures obvious social factors that affect the health and well-being of people with disabilities, inherently eugenic in nature?

3) Does the biomedical construal of "risk" of foetal "abnormality" that genetic counsellors use promote control of women's bodies, and offer spurious projections of the life prospects of people with disabilities?

4) Does the presentation of prenatal screening and testing to detect foetal "abnormalities" as inherently good technology constituting "routine prenatal care" put women under undue pressure to undergo testing to ensure the "normality" of their foetus, effectively reducing their real options with respect to prenatal care, their ability to make fully-informed decisions and their own well-being?

5) Is the increasing institutionalization of genetic screening and testing within standard protocols for prenatal care, and the emergence of genetic counselling as an integral component of health care systems problematic in the absence of guidelines with respect to best practices for genetic counsellors?

## 1.2 Research Activities

The research activities included:

- A literature review of theoretical and scholarly work on prenatal testing and genetic counselling
- A review and analysis of curricula material for pre-service and in-service education and training programs for genetic counsellors
- A review of information resources developed through the Canadian Association of Genetic Counsellors
- A review of relevant case law
- A review of professional and policy regulations
- Interviews with women who have received prenatal genetic counselling
- Focus groups with genetic counsellors, instructors of university genetic counselling programs, medical geneticists, women who have received genetic counselling, and persons with disabilities.

# 2

# SUMMARY OF RESEARCH RESULTS

The concerns that gave rise to this initiative were borne out in the research. Overt eugenic policies such as those in place in Germany under Hitler, and in many other countries, including Canada, during the first half of the 20th century remind us of what can happen if we are not careful and aware. Present day eugenic activity is more covert in nature and has taken many of us unaware. Paid for by the state, protected in law, promoted by the medical establishment as routine prenatal care, sanitised by language that has co-opted the rights discourse of the women's movement, wide-scale identification and (ex)termination of foetuses deemed to be "defective" are being carried out on a scale never before imagined, under the guise of "socially-responsible behaviour" and "informed choice". Given the stated objective of eugenicists such as Frederick Osborn[1] to transform eugenics into a covert activity so that it could win back public favour in the aftermath of Hitler, chilling questions are raised as to the current directions of genetic testing and counselling, medical genetics, and the human genome project itself.

The following quote from a recent statement by the Board of Directors of the American Society of Human Genetics is indicative of the subtle ways in which this new eugenics is justified: if people choose to devalue people with disabilities and engage in what many people with disabilities consider to be genocide – and it's not coerced – then it's not eugenic and is acceptable (responsible) behaviour.

> Galton defined [eugenics] as "the science of improvement of the human race germ plasm through better breeding." At the height of the eugenics movement in the 1920s, the Encyclopedia Britannica (1926) entry on eugenics emphasised that the term connoted a "plan to influence human reproduction." A typical modern dictionary definition is "a science that deals with the improvement (as by control of human mating) of hereditary qualities of race or breed" (Webster's 1983). Although it is not apparent from the dictionary definition, the word has a pejorative connotation, and is frequently used in reference to governmentally driven policies to limit reproductive freedom.
>
> Knowledge-based decisions made by individuals or couples to avoid the birth of a child with disease or disability, so long as they are not unduly influenced by coercive governmental, institutional, or other policies, are acceptable.
>
> Many public health practices to improve the health of living or future people have been implemented to achieve laudable goals. Examples include newborn screening programs to identify infants with disorders for which early treatment is beneficial, the provision of prenatal diagnostic services, maternal vaccinations for rubella, addition of folic acid to food to reduce the risk of certain birth defects, and warnings on alcohol or cigarette labels about the potential damage to the foetus. The American Society of Human Genetics views prenatal screening and diagnostic programs, including those undertaken with the knowledge that an individual who chooses to be tested may seek selective termination of pregnancy, as acceptable as long as individuals are not coerced.[2]

Therein lies the heart of the issue. As the literature review reveals, through euphemisms and cooptation of the language of rights, and through "tactics of confusion,"[3] what once would have been considered as coercion is now presented as "choice". The new eugenics demands that ***individuals*** (as opposed to the State) take responsibility for arbitrating normality. Under this new eugenics, pregnant women must take moral and social responsibility for arbitrating normality, in the context of a genetic counselling process that presents the illusion of autonomous, informed decision-making but within a context that is often neither "value-free" nor "non-directive." The process of drawing women in is so subtle, that most women are unaware that they are unwittingly participating in what can only be described as one of the most successful social experiments in eugenics. Women often do not realise that by receiving "routine" prenatal care such as prenatal ultrasounds and maternal serum screening that they are being committed – often without their consent – to having to arbitrate normalcy.

The evidence presented in this research documents the process through which medical science has been permitted to set the bounds of "normalcy" and how it has systematically excluded people with disabilities from any possibility of being considered normal, of being healthy. It documents the eugenic roots of the narrow biomedical definition of health that is still so firmly entrenched in the medical establishment and in the textbooks used to train medical professionals and genetic counsellors. The geneticisation of medicine further embeds this notion, locating the cause of disability and illness in a person's genes rather than in the social, economic, political, and cultural environments in which a person lives.

This report calls for the removal of the biases that preclude people with disabilities from being considered normal, an end to the tactics of confusion that make informed decision-making impossible, and for the establishment of a regulatory body outside of the medical profession to guide genetic counselling and testing and the setting of bounds for normalcy. It also argues for a broader, holistic definition of health as a state of complete physical, mental, spiritual and social well-being rather than the narrow biomedical definition of health which targets disability and disease for elimination and results in a widespread and systemic devaluation of people with disabilities.

Women themselves often are unable to accept that their child may be "abnormal," or may be unwilling to accept the perceived "burden of care." Research conducted for this project and over the years by *The Roeher Institute* demonstrates how the popularisation of a negative view of disability results in wide-scale intolerance of disability and other deviance from what is perceived to be the "norm." This devaluing of people with disabilities has resulted in a failure to put into place the supports and services that would enable people with disabilities to lead full lives as active members of their families and communities. People with disabilities and their families often experience marginalisation and exclusion, not because of their particular condition per se, but because of the societal barriers to full inclusion. Negative stereotypes and lack of recognition or awareness of the richness of the lives people with disabilities may enjoy when they are well-supported, also help to paint a bleak picture of disability.

Medical professionals and genetic counsellors may themselves perpetuate these perceptions because of their own biases, because they are not well enough informed to challenge them, or because – out of concern for "autonomous" decision-making – they attempt to be "non-directive" in their approach, leaving women in a moral and ethical vacuum.

To be sure, this is not always the case. Many trained genetic counsellors and informed physicians who have had positive experiences with disability make efforts to share these with the women they counsel. Trained genetic counsellors who participated in the research gave evidence that they personally had made sincere efforts to counter the negative images and stereotypes around disability when counselling pregnant women as had others in their profession. Women who had received genetic counselling who were interviewed as part of this research confirmed this.

Unfortunately, trained counsellors do not provide most genetic counselling. Family doctors, obstetricians, and medical geneticists provide much of the initial counselling, and the counselling ability of these various professionals has been found lacking by several studies.[4] Evidence suggests that termination rates may be up to 50 per cent higher when counselling is done by a medical professional rather than by a trained genetic counsellor.[5]

It was also very clear that no matter how well genetic counsellors succeed in trying to present disability in a positive light, the genetic screening, testing, and counselling processes are inherently biased against people with disabilities. Women participate in genetic testing and counselling *because* their foetus already has been labelled as "abnormal" or "defective" or "at risk" of being so. If Down Syndrome, for example, had not been labelled as abnormal and targeted for elimination through prenatal screening programs, pregnant women would be no more likely to go through testing and counselling than if their foetus had a risk of being left-handed, or female – both genetic attributes that also often result in social disadvantage.

Given this inherent bias, a framework for genetic counselling that would value diversity was developed drawing on the literature, and tested in focus groups. It includes a set of shared principles, a model of ethical practice and a set of roles for genetic counsellors. While there was general agreement with the proposed framework during the focus groups, it also became apparent that the underlying structural and ideological issues that frame the current environment in which most genetic counselling occurs must also be addressed. The very real issue to be addressed that emerged from the focus groups and from the literature is: "Does genetic medicine and do genetic counsellors have the capacity to combat the coercive and oppressive incursion into genetics of eugenics?"[6]

This research initiative raises a number of serious issues to be addressed in further work. It points to the huge challenge that lies ahead to bring about a valuing of people with disabilities, in particular those with intellectual disabilities, to reclaim the language of "rights" and "choice" for women and people with disabilities, and to remove the deeply embedded eugenic bias in our society and in the rapidly emerging field of genetics.

## NOTES

[1] See, for example, Osborn (1968).

[2] American Society of Human Genetics (1998).

[3] See Somerville (2001), pp. 119-143.

[4] See Abramsky et al. (2001); also Marteau & Dormandy (2001).

[5] Homles-Seidle et al. (1987); Robinson et al. (1989).

[6] Turnbull (2000), p. 856.

# 3

# BACKGROUND

## 3.1 What is Genetics?

How genetics is understood largely depends on who is defining it. In its purest form, genetics is:

> ..."the study of biological variation." In many ways life can be described as a series of distributional curves. There can be no such things as a "perfect" or even "average" human being...we are all genetically unique. In fact our strengths are our differences. In biological terms variety ensures survival of the species... Where we happen to set the boundaries of what we consider "normal" is very clearly subjective and varies widely in different cultures and eras. This is one of the fatal flaws of eugenics. Since our very essence is our diversity, there can be no single "ideal" to strive towards, even supposing we understood genetics enough to attempt to engineer it.[7]

When genetics enters into the medical arena, the definition becomes much more narrow. For example, "medical genetics" has been defined as:

> ...any application of genetics to medical practice. It thus includes studies of the inheritance of diseases in families, mapping of disease genes to specific locations or chromosomes, analysis of the molecular mechanisms through which genes cause disease, and the diagnosis and treatment of genetic diseases.[8]

Another author has stated that:

> Human genetic variation is associated with many, if not all, human diseases and disabilities, including the common chronic diseases of major public health impact. Because the broad mission of public health is to fulfill society's interest in assuring conditions in which people can be healthy, there will be an unavoidable integration of new genetic technologies and information into public health programs to target intervention strategies that will prevent morbidity, mortality, and disability from a wide array of conditions.[9]

## 3.2 History of Eugenics

Eugenics is defined as "the science of improving the physical and mental qualities of human beings through control of the factors influencing heredity."[10] Eugenics has origins dating to the 19th century and Francis Galton's coinage of the term "eugenics," based upon Charles Darwin's ideas on evolution contained within *The Origin of Species.*[11]

Eugenics was born during a time of social unease in Britain. The British upper and middle classes had successfully restricted their fertility, and there was great fear that society would be overrun by a fast breeding poor working class. In addition, members of the working class were seen as the breeding ground for many social ills, with their wretched lives, chronic illness and physical weakness. The eugenic argument that selected breeding could cure the social ills facing many countries during this period, quickly found favour in Britain, the United States, and in Canada.

It also contributed to the political and social power the medical profession had secured in the Western world by the early 1900s. By the turn of the century, sordid living conditions and contaminated water had been identified as key public health risks. Medical professionals struggled to maintain their social position as responsibility for public health shifted to public works officials and engineers. It is not surprising that medical professionals were among the strongest advocates of eugenics. By situating the cause of social ills in hereditary physical and mental deficiencies and illness embedded in the "germ plasm" of individuals, physicians successful argued that the social ills of the time were in fact medical problems that only they were competent to handle. Dr. Helen MacMurchy, appointed the first chief of the Division of Maternal and Child Welfare in the Ontario Department of Health in 1920, played the most influential role in Canada in securing the supremacy of the medical profession in the public health arena. By firmly establishing the link between eugenics and public health she helped to shift the definition of health to a narrow, individualistic, and biological definition rather than one that recognised the social and environmental causes of ill health.[12]

Significant points of reference in the history of eugenics in the 20th century include:

- Sterilization laws in about half the U.S. states, as well as in several Canadian provinces (the last Sexual Sterilization Acts to be repealed in Canada were in Alberta (1972),[13] and in British Columbia (1973);[14]
- Post-World War II eugenic laws in Japan, activists of which focused on "women and their bodies as a crucial site for successful implementation of eugenics theory;"[15]
- The well-known attempt to create a "superior race" through elimination of "defectives" in Nazi Germany, including "retarded and epileptic children". During the first year of operation of the racial hygiene laws, 80,000 people were sterilised without their consent;[16]
- The passing of a eugenic Immigration Act in Canada in 1910, which prohibited entry into Canada of people deemed mentally defective, including "idiots, imbeciles, feeble-minded, epileptics and the insane, the diseased, and the physically defective, including the dumb, blind or otherwise handicapped."[17]

It has been argued that while the means of eugenics versus "euphenics" (a term coined as an umbrella for such practices as prenatal diagnosis, gene therapy, and therapeutic abortion[18]) differ, the ends remain essentially the same (i.e. the elimination of "defective" genetic traits). While eugenics was situated within the concept of an "ideal race" and the improvement of the population and society as a whole, the procedures of prenatal diagnosis and genetic counselling are situated within a discourse that places highest importance on the prevention of individual suffering, hardship, and risk. Just as the now-taboo science of eugenics was accepted as part of an historical ideology of societal improvement, euphenics has been widely accepted as a natural offshoot of the current neo-liberal theories (advocating individualism and personal autonomy) that serve as the basis for much of modern Western thought and governance.

Hitler's eugenic program to achieve purity of the German race resulted in the much publicised experimentation with and sterilization of people with disabilities, and in the extermination of people he deemed to be deviant – people with disabilities, homosexuals, Catholics, Jews.

Eugenicists in Canada put forward very similar arguments. Dr. William Hutton, medical health officer of Brantford, Ontario and an Executive of the Eugenics Society of Canada (ESC), was a strong proponent of sterilization of the "unfit." During his presidential address to the 1935 meeting of the ESC, he declared "that the Nazis had already put into place an admirable sterilisation law." He conceded that the German law was severe, but in the long run he was confident that it would eliminate those who "clutter up our social institutions."[19]

The extremity of Hitler's measures eventually brought negative publicity to eugenics, and prominent American and English eugenicists who had collaborated with the Germans sought to distance themselves from Hitler's eugenics. Eugenics slowly lost favour outside of Germany, but it did not disappear.

Eugenics in the Post World War II period retained the same goals as before, but assumed a very different look. Armed with the knowledge that covert eugenic activities would not be acceptable, eugenicists sought means to achieve their ends through activities that would gain public approval.

Their belief that widespread acceptance of contraception, sterilization, and abortion were the vehicles to ensure that only the best "stock" would be born, fuelled the birth control movement. It became one of the most potent vehicles for spreading eugenic ideas around the globe, hidden in the language of women's rights. The Planned Parenthood slogan "Every child a wanted child" had its roots in Frederick Osborn's treatise on eugenics, *The Future of Human Heredity*, in which he stated that:

> The most important eugenic policy at this time is to see that birth control is made equally available to all individuals in every class of society…Eugenic goals are most likely attained under a name other than eugenics… Everyone wants children to be wanted children…Proposals based on such criteria are the best we can be sure of at present. They are fully acceptable to the public. Every advance of science will modify and enlarge them."[20]

Osborn was well-placed within the eugenics community, and as secretary to the American Eugenics Society (AES) from 1928 to 1972 held considerable sway. AES members like Margaret Sanger, Dorothy Brush, Frederick Osborn, and Abraham Stone established the American Birth Control League, The International Planned Parenthood Federation and the Population Council to serve their eugenic goals.[21] In her book, *The Pivot of Civilization*, Margaret Sanger outlines the eugenic ideas that lay behind the birth of these organizations:

> Those least fit to carry on the race are increasing most rapidly …Many of the children thus begotten are diseased or feeble-minded; many become criminals. The burden of supporting these unwanted types has to be bourne by the healthy elements of the nation. Funds that should be used to raise the standard of our civilization are diverted to the maintenance of those who should never have been born…The American Birth Control League aims to enlighten and educate all sections of the American public in the various aspects of the dangers of uncontrolled procreation and the imperative necessity of a world program of Birth Control. The League aims to correlate the findings of scientists, statisticians,

> investigators, and social agencies in all fields. To make this possible, it is necessary to organize various departments:
>
> RESEARCH: To collect the findings of scientists, concerning the relation of reckless breeding to the evils of delinquency, defect and dependence;
>
> INVESTIGATION: To derive from these scientifically ascertained facts and figures, conclusions that may aid all public health and social agencies in the study of problems of maternal and infant mortality, child-labor, mental and physical defects and delinquence in relation to the practice of reckless parentage.
>
> HYGIENIC AND PHYSIOLOGICAL instruction by the Medical profession to mothers and potential mothers in harmless and reliable methods of Birth Control in answer to their requests for such knowledge.
>
> STERILIZATION of the insane and feeble-minded and the encouragement of this operation upon those afflicted with inherited or transmissible diseases, with the understanding that sterilization does not deprive the individual of his or her sex expression, but merely renders him incapable of producing children.
>
> EDUCATIONAL: The program of education includes: The enlightenment of the public at large, mainly through the education of leaders of thought and opinion – teachers, ministers, editors and writers – to the moral and scientific soundness of the principles of Birth Control and the imperative necessity of its adoption as the basis of national and racial progress.
>
> POLITICAL AND LEGISLATIVE: To enlist the support and cooperation of legal advisers, statesmen and legislators in effecting the removal of state and federal statutes that encourage dysgenic breeding, increase the sum total of disease, misery and poverty and prevent the establishment of a policy of national health and strength.
>
> ORGANIZATION: To send into the various States of the Union field workers to enlist the support and arouse the interest of the masses, to the importance of Birth Control so that laws may be changed and the establishment of clinics made possible in every State.
>
> INTERNATIONAL: This department aims to cooperate with similar organizations in other countries to study Birth Control in its relations to the world population problem, food supplies, national and racial conflicts, and to urge upon all international bodies organized to promote world peace, the consideration of these aspects of international amity.[22]

Sanger's was a reasoned, scientific approach to the elimination of undesirable traits, in contrast to the emotional, almost hysterical appeal held by Hitler and his fellow eugenicists. It was argued that such a logical, rational approach would help to win back public favour for eugenics. The newly emerging field of "medical genetics" offered the perfect vehicle. In his book, *The Future of Human Genetics*, Frederick Osborn describes the role he foresaw medical genetics playing in the sanitization of eugenics:

> Eugenic proposals had been enacted into law without the scientific evidence to support them. Racial and emotional prejudices had played a part…The old proposals had no solid scientific basis; the newer eugenic policies are based on recent large-scale studies of population trends in this country and on the recent findings of geneticists, sociologists, and psychologists ... A change in attitudes is an essential step toward the development of policies for reducing defects and abnormalities that have a genetic origin…Fortunately the climate for such change is rapidly improving with the advance of knowledge in the field of what is now called "medical genetics"…The new eugenic policies do not give offence.[23]

In Canada, Madge Thurlow Macklin, "the country's pre-eminent human geneticist and the most important scientific defender of eugenics,"[24] took a similar position. In a 1939 journal article, she argued that "eugenic measures should logically be accepted in "a population which already practices birth control extensively for economic and social reasons. Granted birth control, it is difficult to understand why its first object should not be the prevention of inferior children."[25]

As breakthroughs were made in the study of genetics and the knowledge transferred to the field of medicine, medical genetics became one of the primary vehicles for the furtherance of eugenics. Genes were sought for a wide variety of undesirable traits. As these genes were identified pregnant women began to be screened for them. Couched in the language of rights and medical beneficence, eugenics became routinized as standard prenatal care for pregnant women.

> Eugenic pressure starts very humbly, with genetic labelling. In Australia, the Institute of Health and Welfare publishes the "Birth Defect Series." This lists 25 major congenital malformations, and puts together a large range of statistics concerning their occurrence. Most Australian States are not required to keep statistics of terminations of pregnancy for congenital malformations, so notification is incomplete. Genetic labelling such as "major congenital malformation" is an indispensable first stage of eugenic terminations. The range of "major" congenital malformations includes such conditions as anencephalus, microcephalus, and hydrocephalus. It also includes Down Syndrome, Spina Bifida, cleft palate and cleft lip. "Major" congenital malformations are often portrayed as "major" reasons for termination…Eugenic pressure occurs through language and context beyond the act of labelling, and is very often through the medically appropriated language of needs and rights. The similarity of medical language to what is socially approved; the commonality and even banality of these expressions obscures some very great differences. Eugenic pressure appears in the gown of medical beneficence. Eugenic pressure, whenever it is brought to bear, requires scrutiny as to its coercive power, the rationale for it being unleashed, and the greater social consequences. The question to consider is whether there is a similar social intolerance of people with disability of genetic origins as there is a medical intolerance of "major congenital malformations." Coerciveness can be identified when something weak is swept aside.[26]

## 3.3 History of Genetic Counselling

Genetic counselling had its roots in this eugenics movement, initially concentrating on reproductive counselling to couples or individuals designed to ensure that offspring would be of the "best stock".[27] Frederick Osborn referred to the importance of it being "a system of voluntary unconscious selection."[28]

During the 1930s, it was known as "genetic hygiene" or "genetic advice." However, following the public outcry against the eugenic movement as practised by the Nazis in Germany and by others in the United States and elsewhere, and wanting to distance the practice from the 1934 German racial hygiene law, Sheldon Reed coined the more politically-correct term "genetic counselling" in 1947.[29] He realised that this gave it the potential to become widely accepted in a way that would not be possible if eugenic terminology were used. The following quote from Reed in the *History of Genetic Counseling* illustrates this point:

> …my practice of divorcing the two concepts of eugenics and genetic counseling contributed to the rapid growth of genetic counseling. Genetic counseling would have been rejected, in all probability, if it had been presented as a technique of eugenics... In Nazi Germany, an attempt was made to use human genetics for the benefit of the State... in my concept (it) is a type of social work entirely for the benefit of the whole family without direct concern for its effect upon the state or politics.[30]

Genetic counselling in the United States first took place in "heredity clinics" where couples went for information about the risk of having a "defective" child. Osborn described them this way:

> Heredity clinics are the first eugenic proposals that have been adopted in a practical form and accepted by the public. They are run by scientists and their findings are based on scientific knowledge. The word eugenics is not associated with them. The couples who go to them for advice are interested in not having an abnormal child, rather than in the less personal goal of improving the race…It is the function of the heredity clinic, after careful examination of the family record, to advise on the chances of the defect being passed on to the child. Reports from these clinics indicate that the couples are considerably influenced by the information they receive in the clinics, and generally but not always, they are influenced in a eugenic direction. Heredity clinics will become more important with every advance in knowledge of human genetics and with every increase in the education of the public.[31]

## 3.4 Prenatal Diagnosis

Prenatal diagnosis involves a number of screening and testing measures. Prenatal screening is employed to distinguish between a "healthy" foetus and one that has been variously described as "defective," "abnormal," "aberrant," etc. In modern clinical practice it takes several forms, including non-invasive procedures such as screening for maternal age and family history of "abnormalities", Prenatal Ultrasound and Maternal Serum Screening (MSS). When an "abnormality" is suspected, more invasive testing procedures such as Amniocentesis and Chorionic Villus Sampling (CVS) are used to confirm a diagnosis.

> Chorionic Villi Biopsy is a more recently developed technique. Some obstetricians and researchers suggest it will be as commonplace as amniocentesis is at the present time. Some suggest it could replace amniocentesis as the primary diagnostic procedure once its safety has been determined… It consists of removing a sample of the small thread-like projections (chorion villi) which grow in tufts on the chorion. The chorion, part of which

> later develops into the placenta, is the outer embryonic membrane. Because it develops from the fertilized ovum it contains identical genetic material which can be analysed. The procedure was developed in China in the early 70's. Its use is under strict control in Canada at the present time. It is done only at certain hospitals which are taking part in a national test study in which 2,000 women are involved in Canada. As yet, there are no data available on miscarriage or other risks, at least, not in Canada. Although the study is incomplete, the procedure is being offered in some U.S. states and in some European countries as an alternative to amniocentesis. Unlike amniotic fluid, the villi are present early in the pregnancy. The samples can best be taken at about 7 to 12 weeks, perhaps even earlier. The analysis can be done quickly, even overnight if there is a good sample.[32]

Prenatal screening such as prenatal ultrasound is routinely carried out on women over the age of 35 who are considered to have a high risk of having a baby with Down Syndrome and other foetal "abnormalities," on women who may have already given birth to a child with a disability or who have a history of disability in the family. It is presented as a necessary intervention that prepares them for any complications that may arise and so that they can use "positive" prenatal test results as the basis of a decision to abort foetuses that carry mutations associated with disease and/or disability.[33]

Several provinces in Canada have instituted province-wide MSS programs aimed at reducing the number of babies born with Trisomy 15, 18 and 21 (Down Syndrome) and neural tube defects (these programs will be discussed in greater detail later in the report). While women are supposed to give their informed consent for MSS this is not always the case. When presented as part of "routine prenatal care," women may agree to MSS without fully understanding to what they are consenting. One well-educated and informed professional woman interviewed for this research project indicated that she had absolutely no knowledge of being screened before she received the "positive" results of her MSS over the phone at work.

These finding are consistent with research carried out in the United States. One study[34] found that just 19 per cent of 177 patients had received formal genetic counselling before being tested, and that only 17 per cent had given written informed consent. In almost 32 per cent of cases, the physician interpreted the results incorrectly. In a survey of 1,248 obstetricians/gynaecologists,[35] only 14 per cent said that they had received informed consent from their patients before going ahead with genetic testing, and 65 per cent reported that they did not consider themselves well informed about genetics. Another study[36] found that only 20 per cent of health professionals who discussed genetics with patients had formal training in genetics.

## NOTES

[7] Ferreira (d/u).

[8] Jorde et al. (2000), p. 1.

[9] Khoury et al. (2000).

[10] University of California-Davis (2001).

[11] Galton was also an originator (building on the ideas of Adolphe Quetelet) of statistical conceptions of normality (and superiority) which serve as the basis for medical ideals of health.

[12] McLaren (1990), pp. 30-45.

[13] Justice Canada (2000).

[14] Public Guardian and Trustee of British Columbia (2001).

[15] Terazawa et al. (1999).

[16] American Society of Human Genetics (1998).

[17] McLaren (1990), p. 56.

[18] Euphenics is defined by the *Academy Press Dictionary of Science and Technology* as "the improvement of a genotypic problem by treatment during the lifetime of a genetically defective individual." See http://www.harcourt.com/dictionary/def/3/7/0/3/3703300.html .

[19] McLaren (1990), p. 120.

[20] Osborn (1968), pp. 98;105.

[21] See Planned Parenthood (2002); New York University (1997, 1997a); American Philosophical Society (2000).

[22] Sanger (1922/1969), *Appendix - Principles And Aims Of The American Birth Control League.*

[23] Osborn (1968), pp. 86;90;104-105.

[24] McLaren (1990), p. 128.

[25] Ibid., p. 143.

[26] Turnbull (2000), pp. 856-857.

[27] American Society of Human Genetics (1998).

[28] Osborn (1956), pp. 21-22.

[29] Wertz (2000), p. 3.

[30] Reed (1974).

[31] Osborn (1968), pp. 93-94.

[32] Cole (1987), p. 8.

[33] Parens & Asch (1999).

[34] Giardello et al. (1997).

[35] Wilkins-Haug et al. (2000).

[36] Lapham et al. (2000).

# 4

# POLICY, PROCESS, AND PRACTICE OF PRENATAL DIAGNOSIS AND GENETIC COUNSELLING

This review of the practice of prenatal diagnosis and genetic counselling focuses on the various policies – whether governmental, professional, or legal – that have an impact upon 1) personal autonomy and freedom of choice of women and their families during pregnancy, and 2) the general cultural view of people with disabilities. It argues that the present environment is one that is conducive neither to women's informed decision-making during pregnancy, nor to a positive conception of the lives of people with disabilities.

Genetic counselling has been described as "a communication process between health care providers who have expertise in genetics and individuals who are at risk or whose children are at risk of"[37] a genetic condition. Prenatal genetic counselling is genetic counselling that is offered to pregnant women or to women considering pregnancy who are considered to be at risk of having an "affected" foetus. It is offered to women at various stages before and during their pregnancy, but tends to be concentrated during the stages of pregnancy when abortion is considered to be an option.

The responsibility of a genetic counsellor lies in educating their clients about the risks of developing or passing on genetic conditions, and the dialogue that is used in this education is potentially filled with biases regarding those people who already manifest genetic conditions, including people with disabilities.[38]

> Genetic counselling is a profession deeply committed to the furtherance of genetic medicine. Given such a commitment, and imbued with medical authority, genetic counselling not merely describes genetic reality to clients, it *constitutes* genetic reality for them and in so doing radically alters their conceptions of the future.[39]

The official bodies that regulate the profession of genetic counselling acknowledge the emotionally loaded situation faced by those who undergo genetic testing, and have consequently laid out several criteria that genetic counsellors are expected to follow. Knoppers[40] has outlined the following criteria that are considered important issues within the profession:

- Communication and counselling
- Consent and choice
- Control
- Confidentiality
- Competence.

In the context of a discussion concerning the policy and practice of prenatal screening and diagnosis, as well as genetic counselling, the concepts of competence, consent and choice, and communication/counselling are central to the discussion that follows. In theory, the discourse between genetic counsellor (or other health professional) and client/patient rests upon the competence of the counsellor in creating an atmosphere conducive to personal reflection and decision-making on the part of potential parents. In theory, it is this competence that helps to ensure the client's control over the outcome of a genetic counselling session, and that also ensures that a client is able to exercise choice and hence give informed consent to any procedure that may follow counselling.

While this process obviously exists within the dialogue between professional and client, there are other factors that affect a client's – that is, a potential parent's – reasoning and decision-making, and therefore their freedom of choice and ability to make informed decisions. If the present discussion is one of the competences of the professionals entrusted with educating women and families about the facts, realities, and risks of prenatal genetic testing, it must also be a discussion of the environment within which these professionals, and their clients, exist. This environment is constructed by many interweaving factors. These include federal and provincial/territorial government health care priorities and objectives, public and private economic interests, legal regulation and safeguards, and popular public attitudes toward reproduction and disability. Issues that arise within the Canadian context of this review include:

1. The increasing government support of prenatal screening programs such as prenatal ultrasound and Maternal Serum Marker Screening, or MSS, as routine prenatal care;
2. Routine prenatal screening of pregnant women considered to be "at risk" as a part of normal prenatal care and recommendations that this be extended to all pregnant women;
3. A lack of professional norms and standards governing genetic counselling, and the wide variety of health professionals – obstetricians, family physicians, medical geneticists, genetic counsellors – involved in prenatal care for whom differing sets of norms may apply;
4. The legal necessity of offering prenatal screening and diagnosis to all pregnant women in light of wrongful birth lawsuits against health professionals;
5. A general lack of public, government-sponsored supports to people with disabilities and their families.

## 4.1 Regulatory Positions of Professional Genetic Counselling Organizations

A number of professional bodies have an influence on the practice of genetic counselling in Canada. These include the American Board of Genetic Counseling, the National Society of Genetic Counselors, and the Canadian Association of Genetic Counsellors.

The American Board of Genetic Counseling (ABGC)[41] describes a genetic counsellor as an individual who provides information about the risk of occurrence of genetic conditions. A counsellor is a medical professional responsible for communicating genetic, medical, and technical information in a manner that is:

- Comprehensive
- Understandable
- Non-directive.

It has been written that, "exploring feelings (*about the birth of a child with a genetic anomaly)* may be far more important than providing a statistical estimate of risk, and somewhere during the counselling process there should be an opportunity to do so."[42] Barr expands on this by pointing out the necessity of balanced, complete, non-directive information, put forward in a manner that conforms to the level of understanding of clients.[43]

According to the National Society of Genetic Counsellors (NSGC), genetic counsellors must:

- Recognise the limits of their own knowledge, expertise, and competence;
- Enable their clients to make informed, independent decisions by providing necessary facts and clarifying relevant issues;
- Be aware of social developments that may endanger the physical and psychological health of individuals.[44]

The Canadian Association of Genetic Counsellors is a body similar to the ABGC and the NSGC in the United States, and holds similar tenets. It states that the duties of a genetic counsellor are to:

- Understand the medical facts related to diagnosis, probable course, and management of a genetic condition;
- Have knowledge of heritable properties and risk of recurrence of a condition;
- Explain tests that are available and coordinate or facilitate genetic testing;
- Understand methods of dealing with the risk of recurrence;
- Choose the course of action that seems appropriate to them in view of their risk, their family goals, and their ethical and religious standards, and to act in accordance with that decision;
- Make the best possible adjustment to the disorder in an affected member and/or to the risk of recurrence of that disorder.[45]

Official representations of genetic counselling (based strongly upon the ideals of informed decision-making) seem to take a positive view of disability, and some go so far as to place emphasis on the idea that it is part of a genetic counsellor's job description to advocate on behalf of people with disabilities. However, not all genetic counselling is provided by a licensed genetic counsellor, especially in less-populated regions or in those that do not have access to teaching and research hospitals.[46]

According to information collected during this research from women who had received prenatal genetic counselling from medical geneticists and genetic counsellors, counselling takes on different forms depending on who is doing the counselling. In reality, approaches range from very directive to non-directive. Some medical professionals take on the responsibility of active encouragement of termination of a pregnancy thought to be at "high risk" of producing a "defective" or "abnormal" baby. One medical geneticist interviewed as part of this research is a senior professional conducting genetic counselling at a large Canadian hospital. This person explained that they actively promoted abortion in certain situations. If there was a risk that a foetus would be affected by a condition such as epilepsy, they would advise against abortion, given the ability to control the condition through medication and the uncertainty as to what degree the foetus would be affected. However, if that condition were coupled with a high risk of an intellectual disability, that person would push hard for abortion, given the burden they perceived this would place on the family. In their view, since couples are choosing to have only one or two children today, as opposed to the large families of the past, they should not have to settle for a "defective" child.

One woman interviewed as part of this research received prenatal genetic counselling following a "positive" MSS result. The counselling happened in a large urban hospital in a team setting. Before seeing the trained genetic counsellor, who she found to be very supportive, she was first seen by an obstetrician who was part of the team. During three visits scheduled over a five-week period the doctor pressured her to have amniocentesis even though she had already refused it. She was told that they had to hurry and have the testing done because the option to abort was there only until 21 weeks into the pregnancy. A series of ultrasounds was done, 17 in all over a six-month period. She found the doctor to be very cold and felt uncomfortable being in the same room with her. She described the doctor's language as "to the point – live with it or get rid of it [the foetus]." She finally refused to be seen by the doctor. The day after she gave birth to her baby who has Down Syndrome, the obstetrician barged into her room and said to her: "I told you that you had a choice, and *this* is what you chose. Live with it." The message she got from the doctor was that "you brought another one of these kids into the world. Why would you do this?"

Another woman interviewed recalled receiving a call from her doctor saying that the baby had a genetic abnormality and may not survive and she may want to consider an abortion. She was told to show up for genetic counselling the next day. She describes being terrified and not knowing what to expect – did the baby have "two heads"? She had no choice but to go for counselling. During the first visit with the trained genetic counsellor, she and her husband were informed that they had a high risk of having a baby with Trisomy 21 and that they had three choices: have an abortion, have amniocentesis, or carry the baby to term. She said that they were shocked that abortion was the first choice offered on the basis of a positive MSS result, without any confirmation that there really was an "abnormality." During that first visit, the counsellor provided them with all kinds of negative information about the risks of the baby not making it to term and the health risks if it did. When they informed the counsellor that they had already made a decision to carry the baby to term, from then on the counsellor "lightened up" and was extremely supportive and helped them to link with resources about Down Syndrome. The mother wondered what parents who were not as well informed and educated as she and her husband, or who had not made up their minds before attending genetic counselling would have done when faced with the information they were given during their first counselling session.

A widely-quoted study found that 67 per cent of couples who received positive genetic information (that is, were informed that their child was at risk of developing a genetic condition) from a family doctor (not necessarily trained in genetic counselling) chose to terminate pregnancy, compared to 39 per cent of couples who received the information from a trained genetic counsellor.[47] This would appear to signal a significant difference in practice between general practitioners and specialists. Questions then arise as to what differences exist between different types of practitioners, why training in genetic counselling causes such a variance, and why almost 40 per cent of couples counselled by a trained genetic counsellor still chose to terminate pregnancy.

Genetic-related knowledge on the part of physicians can be less than perfect.[48] Another relevant point is that since different health professionals are regulated by different professional bodies, each are not bound by the standards of the others. Thus, all professionals who actively counsel patients regarding prenatal diagnosis do not necessarily adhere to the tenets held, for example, by the Society of Obstetricians and Gynaecologists of Canada (SOGC) and the Canadian College of Medical Geneticists (CCMG).[49]

A 1994-95 study found that sessions involving genetic counsellors focus for the most part not on the exploration of feelings, nor on the psychosocial issues related to raising a child with a disability, but instead on diagnosis and the estimation of risk.[50] This is problematic, as the concept of risk is one that may be poorly understood by potential parents. It has been found that clients tend to overestimate risk figures – for example, a risk estimate of 3 per cent (low by most standards) may be interpreted as moderate, and a risk estimate of 25 per cent (moderate by most standards) is likely to be interpreted as high to very high.

"Changes in risk perception also may result from the increased availability of prenatal diagnosis and other reproductive options that allow people to interpret their risks as high, because they may have a possibility of averting the risk."[51] One woman interviewed for this project told how, after giving birth to a baby with Down Syndrome, she was advised by a genetic counsellor that on the basis of what was described to her as a 1 per cent risk factor, she had a high risk of having another baby with Down Syndrome. She decided against another pregnancy. On the opposite side of the equation, in areas where prenatal diagnosis is not widely available, risk estimates normally considered high may be minimised.

## 4.2 Current Regulation in Selected Provinces

The current approach to the regulation and monitoring of genetic counselling in Canada could arguably fall under what Knoppers terms the "private policy approach."[52] This approach unfolds in the following steps:

1. Knowledge is collected and practice established through clinical experience and empirical study;
2. Professional practice guidelines are developed based on the consensus of professionals experienced in the field;
3. Regulation is developed at the "techno-scientific level,"[53] and may lead to legislation.

This state of affairs is relevant to the discussion of the relationship between professional clinicians, the services they provide, and the potential conflict that arises when groups of individuals are responsible for the regulation of the very thing that provides their livelihood – as Turnbull writes, "genetic counselling is a profession deeply committed to the furtherance of genetic medicine."[54] To set the stage for a discussion of implications in a Canadian context, as well as to illustrate the general lack of legislation concerning the proliferation of prenatal testing, following are examples of the current state of genetic counselling regulation in selected Canadian provinces.

### *British Columbia*

The British Columbia Ministry of Health and Ministry Responsible for Seniors has published guidelines regarding clinical prenatal testing, which state:

> When a foetal abnormality which is associated with an increased risk of foetal chromosomal abnormality is detected on a prenatal ultrasound and is the only indication for prenatal cytogenetic testing, consultation with a certified clinical geneticist or maternal-foetal medicine specialist is mandatory. When the clinical geneticist or maternal-foetal medicine specialist determines that the risk of a foetal chromosomal anomaly is equal or greater than 0.5 per cent, then prenatal cytogenetic testing should be offered.[55]

Nowhere is it mentioned that a consultation with a genetic counsellor should be offered before or after the offer of genetic testing. Counselling is also not listed in the British Columbia Medical Services Commission Payment Schedule with regard to any form of prenatal testing (e.g. chorionic villus sampling, amniocentesis etc.).[56]

## Ontario

The following instructions are offered in the Obstetrics section of the *Ontario Health Insurance Schedule of Benefits and Fees: Schedule of Benefits for Physician Services*:

> A prenatal major assessment includes a full history, and an examination of all parts or systems (and may include a detailed examination of one or more parts or systems), an appropriate record and advice to the patient. All other prenatal visits include the necessary history, examination, appropriate record and advice to the patient. All prenatal visits (major and minor), include pregnancy-related counselling as a form of providing advice to the patient or the patient's representative(s).[57]

The alphabetic index of the Schedule of Benefits does not specifically list genetic counselling as a fee-for-service.[58] The "Consultations and Visits" section lists "Genetic Family Counselling" as:

> ...counselling dedicated to an educational dialogue between the physician and one or more family members, guardians of a genetic patient or person authorised to make a treatment decision for the genetic patient for the purpose of providing information regarding treatment options and prognosis.[59]

While this does indicate the necessity of genetic counselling, it would seem that this is directed not towards pregnant mothers, but instead toward those people who choose to be tested for conditions such as breast cancer and Huntington's disease, among others that have a genetic component. These accounts suggest that, at least according to medical regulation, there is substantial leeway for a family physician to make decisions according to preference, rather than according to a standardized protocol.

## Newfoundland

In Newfoundland, there is at the present time no legislation regulating the provision of genetic counselling with reference to prenatal testing. A recent government document has expressed concern with "research done by people coming into the province and studying the DNA of families without following up with these families as to the outcomes of the studies."[60] This refers to a situation commonly found in areas whose population has descended from a small number of settlers (and therefore a limited gene pool) and has not been expanded by subsequent high rates of immigration. This is also the case in the northern Quebec region of Saguenay/Lac St. Jean, where certain genetic conditions (such as Spastic Ataxia) have been found in higher rates than would be expected in a more diverse community. The document goes on to say that "for this reason in particular, it is important that the province move forward with policies and standards regarding all genetic research in Newfoundland and Labrador."[61] A report was commissioned by Newfoundland Health and Community Services to examine the issues raised by the current research undertaken in the province.

## 4.3 Regulation versus Practice in Genetic Counselling

A number of researchers have drawn distinctions between what is considered to be best practice in genetic counselling and the actual practice of genetic counselling in Canada. For example, Burgess et al. have examined the necessity of proper genetic counselling on one hand, and the reality of genetic counselling on the other. Their recommendations are similar to the tenets put forth by the various Canadian and U.S. bodies that oversee the profession (i.e. the NSGC, SOGC, CCMG, CAGC). In their view, genetic counsellors should have up-to-date knowledge of genetics and genetic testing, must be aware of the psychological and social factors involved in testing, and must be able to provide clinical follow-up to their clients.[62]

Outside of ethical ramifications (for example, failure to ensure complete information disclosure and thus not fulfill obligations of informed consent) and civil actions regarding wrongful birth, medical negligence, etc., it appears that medical professionals, whether family physicians, obstetricians, or trained genetic counsellors, are left to provide prenatal genetic counselling in a largely unregulated environment. They find themselves in second place in a technological hierarchy, at the top of which are genomic researchers and companies who fund the development of genetic testing. Professional and regulatory bodies struggle behind to understand the psychological and emotional effects that prenatal genetic diagnosis and counselling may have on those who receive it.

While prenatal genetic diagnosis is going ahead full steam in Canada, there are no provisions in place to ensure that women offered genetic screening and testing are receiving genetic counselling, nor that when they do receive counselling it is from certified counsellors knowledgeable about genetics, the inherent value of people with disabilities, and of the important contributions people with disabilities make to families and communities. Burgess et al. point out the shortage of professionally-trained genetic counsellors and clinical geneticists required to handle the demand for increasingly commonplace and in-demand prenatal genetic diagnosis – as of September, 2001, there were 189 genetic counsellors registered with either the CAGC or the NSGC practising in Canada.[63]

While authors have highlighted the importance of the presence of competent counselling professionals, they also direct attention to studies which reveal that "among Canadian health care professionals, understanding of genetics is poor, and there is wide practice variation with respect to genetic testing."[64] And while many patients may be referred for a consultation with a trained genetic counsellor, "it is inevitable that physicians will be involved in long-term follow-up both by monitoring for disease and by supporting the integration of genetic information into patients' lives."[65]

It has also been suggested that although the ethos of genetic counselling places great emphasis on a non-directive, non-biased presentation of available options in a genetic counselling encounter, "the subtle influence of counsellors' values may affect patients' choices."[66] Of similar relevance is physicians' choice to refer patients to a genetic centre or take on the responsibility of counselling themselves. Differences in abortion rates of women counselled by genetic counsellors versus women counselled by family physicians (39 versus 67 per cent, respectively[67]) raises the question of whether medical professionals not trained as genetic counsellors are providing sufficient information to ensure that women (and all family members involved in pregnancy-related decision-making) who choose termination of a foetus do so on an informed and autonomous basis.

## 4.4 Regulation through Civil Action – Wrongful Birth Legal Action

A recent Canadian lawsuit against two family physicians who practice obstetrics resulted in the award of $3 million to the parents of two children who have Muscular Dystrophy. The parents sued because of a "legal regret that the children had been born."[68] The suit was won when it was decided that the physicians failed to meet the current standard of medical care, as they did not refer the prospective parents for prenatal genetic counselling – if the parents had known they were at risk, they would have terminated the pregnancies.

This situation is illustrative of two major points – first, that prenatal diagnosis and genetic counselling, while not strictly regulated or controlled, are considered to be a necessary aspect of the current standard of prenatal care. While the law requires a doctor to offer any tests or treatments reasonably indicated, it does not give the right to impose any treatment or test. Nonetheless, some women are tested for genetic anomalies without their consent, and many are made to feel that they are "socially irresponsible" if they fail to submit to such testing. The second point concerns the decision that the parents in the situation described above would have been justified in choosing abortion in the case of a risk of disability. This case is not the only one in which damages have been awarded to parents who were not referred for genetic counselling by their physician, or where the doctor failed to present a clear picture of the extent of "risk of abnormality" involved.[69]

As Caulfield and Robertson write, "the mere existence of wrongful birth and wrongful life claims may cause physicians to encourage the use of genetic services and to subtly recommend the termination of pregnancies that may result in the birth of a child with a disability."[70]

## 4.5 Government Promotion of Genetic Screening – the Maternal Serum Screening (MSS) Programs in Ontario and Manitoba

The assertion that prenatal testing is becoming increasingly institutionalised within what is considered "normal" prenatal care[71] is borne out in the presence of government support and widespread proliferation of Maternal Serum Screening in two Canadian provinces. Under the program in Ontario, it is recommended that all pregnant women be offered testing, "with counselling about the test, possible outcomes, options for further testing, and risks."[72] It was found that 88 per cent of respondents to a recent study (including family physicians, obstetricians, and midwives currently providing antenatal care or attending births) offered the test to all pregnant women on a routine basis.[73]

The program also exists in Manitoba, though it would appear that the test is not offered on as widespread a basis as it is in Ontario – in 1992, it was found that 60 per cent of survey respondents (of the same professions as those in the Ontario study) offered the test to all pregnant women.[74] More relevant to this discussion is the fact that 22 per cent of these indicated that the test was performed without the consent of the patient, as a segment of routine prenatal blood work.[75]

The evaluations of the program in Ontario shed light on some practical issues related to Maternal Serum Screening. First of all, as is the norm within the medical community, the program is largely regulated by the same professionals who administer it. There is little regulation in place that would prevent a physician with little knowledge of genetics, or of MSS itself, to discuss it with patients. This fact takes on added relevance considering that, in a survey administered in one study, one-third of respondents did not even complete questions related to knowledge of issues around MSS.[76] Professionals who did answer these questions grossly overestimated the accuracy of MSS – while only 2 to 3 per cent of women with positive MSS results will actually have babies with a Neural Tube Defect or Down Syndrome, the average professional who administers the test believes, on average, that this figure is actually almost 16 per cent.[77]

According to a recent report, the situation in Ontario is similar to that in British Columbia. Availability of testing is limited outside of the Vancouver and Victoria areas. Responsibility for screening and counselling has been placed in the hands of physicians and obstetricians in the absence of a proliferation of trained genetic specialists. There is no body in place to oversee and regulate standards and funding, and there is no arena within which issues relevant to individuals with disabilities and their families can be heard and discussed.[78] This illustrates the discrepancy between funds accorded to screening and those provided to supporting individuals and families.

While the British Columbia study found wide support for a coordinated, government-funded screening program (which has not been endorsed or initiated by government itself), the reports from Ontario suggest that the reasons for this support may be unrealistic. These reasons include:

- The present system of uncoordinated, "ad hoc" funding has resulted in unequal access to screening services for those outside urban areas;
- Lacking coordination, there is no system for dissemination of knowledge to physicians, obstetricians, and the general population;
- Lacking coordination, there is no system for dissemination of guidelines and standards of practice.

Supporters of province-wide MSS testing in British Columbia argue that provincial coordination of screening would improve practice, expand general knowledge of issues surrounding disability and prenatal diagnosis, and predicate greater equality of access to screening services. However, the findings of studies in Ontario cast doubt on whether this would actually be the case. Judging from evaluations that took place in 1997-99, it can be said that high levels of practitioner knowledge, equality of access, and standardization of care have not come as a result of centralized "coordination." Stemming from concerns about effective counselling and support services, as well as a perceived lack of social and cultural sensitivity of the screening program, practitioners in Northern Ontario were less likely to offer MSS, and were more critical of the program, than were those in urban centres.[79] And as discussed above, practitioner knowledge concerning MSS within this "coordinated" system is far from optimal.[80]

## NOTES

[37] Sidarous & Lamothe (1995), p. 154.

[38] See Vehmas (1999); Parens & Asch (1999); Diesfield (1999).

[39] Turnbull (2000), p. 854.

[40] Knoppers (1995).

[41] American Board of Genetic Counselors (1996).

[42] Fraser (1974), as quoted in Barr (1999), p. 31.

[43] See Barr (1999).

[44] National Society of Genetic Counselors (1995).

[45] Canadian Association of Genetic Counsellors (2001).

[46] Sidarous & Lamothe (1995).

[47] Robinson et al. (1989).

[48] See Sidarous & Lamothe (1995); Wertz (2000).

[49] Wertz (2000).

[50] Wertz (2000).

[51] Wertz (2000), p. 15.

[52] Knoppers (1995).

[53] Knoppers (1995), p. 3.

[54] Turnbull (2000), p. 854.

[55] British Columbia Ministry of Health and Ministry Responsible for Seniors (2000).

[56] British Columbia Medical Services Commission (2001).

[57] Ontario Ministry of Health and Long Term Care (2001a).

[58] Ontario Ministry of Health and Long Term Care (2001b).

[59] Ontario Ministry of Health and Long Term Care (2001c).

[60] Newfoundland and Labrador Health and Community Services (2000).

[61] Ibid.

[62] Burgess et al. (1998).

[63] Information generated through private consultations with the Canadian Association of Genetic Counsellors and the National Society of Genetic Counsellors, September 2001.

[64] Burgess et al. (1998); see also Wertz (1995); Laberge et al. (1995).

[65] Burgess et al. (1998).

[66] Ibid.

[67] Robinson et al. (1989).

[68] Cohen (1999).

[69] For more examples, please refer to section 6.4 of this report.

[70] Caulfield & Robertson (1996), p. 59.

[71] See Browner & Press (1995).

[72] Carroll et al. (1997).

[73] Ibid.

[74] Chodirker & Evans (1993).

[75] Ibid.

[76] Carroll et al. (1997), p. 776.

[77] Ibid., p. 783.

[78] British Columbia Office of Health Technology Assessment (2000).

[79] Permaul-Woods et al. (1999).

[80] Carroll et al. (1997).

# 5

# TRAINING OF GENETIC COUNSELLORS

Despite any emphasis that training programs for genetic counsellors may place on understanding the lives of people with disabilities and their families and on non-directive approaches to counselling, the training on genetics is biased against people with disabilities. As the following analysis of various books used in university training programs on genetic counselling in Canada reveals, students are trained from the outset to view disability as "abnormal," a "defect," and something "undesirable" for both persons with disabilities and their families. With a shift in the focus of medical genetics, and a concomitant shift in the training of genetic counsellors away from these biased materials, it is possible that the bias against people with disabilities inherent in genetic counselling can be removed. The conditions needed for such a shift to occur need to be fully explored and communicated.

## 5.1 Content Analysis of Genetic Counselling Curricula Materials

The literature review undertaken for this research analysed a number of curriculum materials used in the three major Canadian Master's level genetic counselling programs. The materials are critically examined from a perspective founded in feminist and disability theory, a perspective represented in the work of authors such as Michael Oliver, Simi Linton, and Adrienne Asch. The review synthesized literature that is based upon a medicalized ideology wherein disability is equated with abnormality, individual pathology, personal suffering, and familial and societal burden. The various course materials hold a globally negative and devaluing position regarding disability and genetic difference. This position may serve to promote the avoidance of the birth of children with genetic differences, as well as the medical invalidation of women's personal bodily knowledge and decision-making, particularly in terms of reproduction.

This review is relevant to two main groups, the first consisting of members of the following professions:

- Genetic researchers and practitioners
- Genetic counsellors
- Family physicians
- Obstetricians and gynaecologists
- Obstetrical and genetic nurses and support workers.

The second group of stakeholders includes people with disabilities and their families, as well as disability advocates and activists. These groups represent two (potentially) opposing points of view: the advancement of scientific and medical technology for the perceived benefit of society, versus the maintenance of human rights and proper ethical boundaries in medical care.

There is an intersection of theoretical constructs within the materials outlined below that illustrates a paradox within the medical/genetic professional community. On one hand, disability is portrayed in a variety of negative and devaluing ways:

- as disease
- as abnormality
- as the cause of poor life prospects
- as a cause of suffering
- as a familial and societal burden.

On the other hand, health professionals are required to be non-directive when counselling pregnant women who may be at risk of giving birth to a child with a disability.[81] There is evidence that this may produce substantial conflict. Regarding foetuses with genetic "anomalies", researchers have found a degree of acceptance of abortion among genetics professionals that "may be disconcerting to the disability community or to patients with these conditions."[82] Researchers have also found a "significant relationship between professionals' personal attitudes and the direction of their counselling."[83] In a 1985 study, 25 per cent of obstetricians surveyed indicated that they provided Maternal Serum Alpha-Fetoprotein (MSAFP) screening "without offering women any explanation of its purpose or any choice about whether or not they participated in the screening program."[84] It was also found that obstetricians were biased towards the uptake of prenatal testing, and the use of abortion services. Marteau found that some health professionals actively attempted to persuade women to undergo prenatal diagnostic testing.[85] Each of these findings is supportive of the widely-held views that 1) genetics professionals are biased towards the use of services related to their professions (including prenatal diagnosis and abortion), and 2) it is impossible, considering the environment within which genetic services exist, for genetics professionals – even those aware of disability issues – to be truly non-directive when offering their services.[86]

The review/content analysis that follows has been carried out with these issues providing direction. This analysis is, at its most basic, an analysis of language. The language in question comes from a point of view that both creates and is created by a hierarchy that places knowledge and intelligence in a position of power over those it concerns. In this instance, those whom it concerns are people with disabilities and, through association, women who are or who plan to become pregnant.

Cocks has written of the idea that members of society who hold greater amounts of power "ascribe status to those below them and influence their degree of participation in society."[87] This status is conferred through language, and as Linton states,

> …language that conveys passivity and victimization reinforces certain stereotypes when applied to disabled people. Some of the stereotypes that are particularly entrenched are that people with disabilities are more dependent, childlike, passive, sensitive, and miserable and are less competent than people who do not have disabilities. Much of the language used to depict disabled people relates the lack of control to the perceived incapacities, and implies that sadness and misery are the product of the disabling condition.[88]

From another perspective, a correlation between suffering and disability may be attributed not to the presence of a disabling condition, but instead to the presence of an environment and social structure that do not reflect the needs of people with disabilities.[89] Herein lies the difference between the medical and social models of disability that the materials reviewed below do not take into account. Swain & French refer to the "affirmation model" of disability, "a non-tragic view of disability and impairment which encompasses positive social identities, both individual and collective, for disabled people grounded in the benefits of life style and life experience of being impaired and disabled."[90]

It is from this perspective that the HRDC Office for Disability Issues has produced guidelines for print and broadcast media professionals regarding the portrayal of people with disabilities. In the introduction, it is stated that:

> ...language is a powerful and important tool in shaping ideas, perceptions, and ultimately, public attitudes. Words are a mirror of society's attitudes and perceptions. Attitudes can be the most difficult barrier persons with disabilities must face in gaining full integration, acceptance and participation in society... demeaning, belittling or negative words are a barrier to greater understanding and can trivialize genuine support given by a community to persons with disabilities.[91]

Master's level genetic counselling course materials were examined according to guidelines such as those provided by the Office for Disability Issues. The materials were divided into three major categories: 1) literature explaining the structure and function of genetic material;[92] 2) literature that analyzes genetic development from a medical perspective, and that focuses on the nature of the developing human in terms of normality and abnormality;[93] and 3) literature that focuses on ethical issues relevant to medicine and the use of genetic technology.[94] Also in use are books focusing on the lives of people with disabilities and their families.[95] This review will deal mainly with the second and third categories – medical genetics and medical ethics.

A notable similarity between resources in these two categories – medical/genetic and ethical/moral – lies in the use of the language of medical genetics. The use of devaluing terms such as *birth defect*, *abnormality*, and *congenital malformation* is widespread. In addition to resources outlined in program book lists and found in faculty libraries, genetic counsellors are informed by journals that also make use of such language. These include *The American Journal of Medical Genetics; The American Journal of Human Genetics; Teratology: The Journal of Abnormal Development;* and *Clinical Dysmorphology*.

Following is an in-depth examination of several resources used in the genetic counselling programs offered by Canadian universities. These are Masters level science programs, and entrants are required to hold Honours Bachelor of Science degrees, with previous education and training in genetics, biochemistry, and statistics. Experience in a counselling setting is also expected. The course load is slightly dominated by courses in genetics and biology (complemented by courses in counselling and ethics), and students are required to participate in clinical rotations. The resources discussed below would be introduced in the first half of the program, when course work is emphasized in order to prepare students for more practical applications.

## 5.2 Analysis of Genetic Counselling Curricula

### *I. Negative Language and Devaluation of People with Disabilities*

*The Developing Human: Clinically Oriented Embryology*[96] is an example of a clinical volume of work that is used to inform genetic counsellors of "normal" human development, and the kind of congenital "anomalies" with which they will come into contact. Contained within the text is a chapter entitled "Human Birth Defects," characterized by language employing such terms as:

- defect
- disease
- mutant, mutation
- abnormal development
- mentally retarded
- mental defectives
- disruption
- deformation
- malformation.[97]

Another text used in a Canadian genetic counselling program is that by Marteau and Slack.[98] The authors show a clear bias toward people with disabilities in their discussion about parents expecting a child "at risk" of having a disability:

> For the minority (of parents) who will receive a positive diagnosis indicating abnormality, it provides the potential to avoid the birth of an abnormal child. At the same time it causes distress and grief from rejecting an unwanted child.[99]

This quote is ideologically similar to comments made elsewhere by Marteau, for example in the preface to Marteau and Richards,[100] in which the authors write of individuals who are "condemned to a life sentence in the knowledge that they, or their children, will suffer from an incurable genetic disease." In the same preface, prenatal diagnosis, selective abortion, and gene therapy are mentioned in the first paragraph. Throughout the preface, various references are made to the "burden of a genetic disorder,"[101] and living in the "shadow of genetic disease."[102]

A resource used in at least two Canadian genetic counselling programs is that by Nora and Fraser,[103] which is prefaced by the authors' "hope that this book will continue to find its place in the classroom, the clinic, and the counselling centre."[104] It is characterized by similar language to that used in the volume by Moore and Persaud – genetic variation is described in terms of "disease," "abnormality," "malformation," "development" versus "maldevelopment," etc. Within the current debate surrounding the ethical necessity of non-directiveness in genetic counselling, the chapter entitled "Prenatal Diagnosis of Genetic Disease" is instructive. The position taken by the authors makes it clear that the potential for genetic counselling to result in the termination of an "affected foetus" is considered to be an exciting source of progress in the field:

> One of the most exciting chapters in the history of genetics as applied to medicine is the development of techniques for the diagnosis of congenital disease before birth. Before this, genetic counselling could provide only probabilities of recurrence, based on the Mendelian laws or empirical data, and couples at risk for such diseases could never have a child without taking the chance that it might be affected. Now it is possible, for certain diseases, to provide a definite answer – yes or no. If the foetus is affected, the parents have the choice of terminating the pregnancy and beginning again.[105]

The chapter is peppered with similar phrases, such as the following regarding amniocentesis:

> ...the longer one waits after this (i.e. 16 weeks after the beginning of the last menstrual

> period), the less time there is to grow the cells, make the appropriate tests, and obtain a diagnosis while there is still time to perform an abortion. Because cultures occasionally fail, requiring another tap, and for some biochemical tests it takes 3 to 6 weeks before the results are known, the schedule may be tight.[106]

Another sentence states: "There may be difficulties in deciding whether minor variations in chromosome morphology are significant abnormalities or normal variants."[107] Rather than questioning why conditions that may result in a disability are not still referred to as "variants," the authors send a clear message that these conditions are in fact "abnormalities" to be eliminated.

In a discussion of Chorionic Villus Sampling (CVS), Nora and Fraser write that "earlier diagnosis has several advantages. The procedure can be done while the pregnancy is still 'private,' and, if an abnormality is found, abortion is simpler, less stressful, and less dangerous than it is at 20 weeks."[108] The authors continue by saying: "On the other hand, the new technique raises more ethical problems, such as pressure to do the test for increasingly trivial reasons and, in particular, for the determination of sex for social, rather than medical reasons."[109]

Failure to question the arbitrariness of this distinction between "medical" and "social" reasons, promotes bias against disability. In fact, many foetal differences considered to be "defects" (not only those deemed "trivial" in the chapter) are stigmatized for social rather than medical reasons.[110] In a related vein, Hubbard equates testing for disability with testing for skin colour. She writes that

> ...most of us would be horrified if a scientist offered to develop a test to diagnose skin colour prenatally so as to enable racially mixed people... to have light skinned children. And if the scientist explained that because it is difficult to grow up black in America, he or she wanted to spare people suffering because of the colour of their skin, we would counter that it is irresponsible to use scientific means to reinforce racial prejudices. Yet we see nothing wrong, and indeed hail as progress, tests that enable us to try to avoid having children who have disabilities or are said to have a tendency to acquire a specific disease or disability later in life.[111]

In Nora and Fraser's discussion of the various prenatal diagnostic techniques (e.g. ultrasound, CVS, amniocentesis), most are judged according to whether or not they are able to diagnose genetic anomalies before the end of the second trimester. In other words, a test is deemed less useful if it is unable to enable diagnosis before the time within which an abortion may be performed. For example, amniography is given little space in the text because "unfortunately, the technique does not seem to be reliable in the second trimester, perhaps because the vernix (*i.e. the fatty layer which covers the fetal skin and which picks up a dye which is injected into the womb)* is not sufficiently developed by then."[112]

While Nora and Fraser write, "that electing for prenatal diagnosis implies a possible induced abortion raises some difficult ethical and moral questions,"[113] they continue to promote a negative view of disability. The following excerpt is illustrative of the overall position of the text toward disability:

> Couples ... should feel free to make an uninfluenced choice to decline amniocentesis and take their chances, and may very well do this if they find that the risks are in the less than 5% range, particularly if they object to abortion on moral or emotional grounds. Or they may accept amniocentesis and abortion if the diagnosis is positive. Or, if they object to abortion, they may accept amniocentesis with the intention of continuing the pregnancy even if the diagnosis is positive. They may want the chance to prepare themselves for care of a defective child either at home or in a foster home or institution.[114]

The authors succeed in laying out the options involved around prenatal diagnosis, but suggest a bias by using the phrase "defective child." Also of note is the reference to foster family and institutional care. The portrait given of life with a child with a disability is overwhelmingly negative. And while the language of the chapter is typical, there is deeper meaning to be found within less obviously negative terms. For example, at one point in the chapter it is said that amniocentesis is used, among other things, to detect "fetal distress."[115] Further on, similar but more obvious phrasing is instructive: "The development of prenatal diagnosis has involved the convergence of many disciplines, ranging from molecular genetics to biomedical instrumentation, on the common goal of reducing human suffering."[116] Here the authors are able to define prenatal diagnosis as something inherently good, in that it is able to prevent human suffering. This judgment is dependent on a characterization of disability as something that universally causes suffering and distress, and of suffering as inherently bad.

Another text used in a major genetics counselling program in Canada is *Principles of Medical Genetics*.[117] The chapter entitled "Ethical Considerations" sends mixed messages – for example, the authors discuss the following ethical principles:

- Respect for autonomy
- Beneficence and nonmaleficence
- Privacy and confidentiality
- Justice and equity.

During a discussion of ethical considerations regarding prenatal diagnosis, the authors draw a potentially important distinction between two types of disability. In a discussion of two parents of a young child with a mild learning disability, the authors estimate a 7-8 per cent chance of having a second child with a similar disability. The authors take issue with the fact that the parents requested a referral for abortion during the second pregnancy. The mother is quoted as saying that having another child with a similar disability would be too great a burden for her. Though the request for referral is granted, the couple is also referred for follow-up counselling to deal with issues that exist concerning their daughter, and it is stated that "every medical geneticist has encountered a case such as this one, where a relatively low risk for a relatively mild condition is viewed by the couple facing that situation as much more significant."[118]

This situation is contrasted with that of another couple, both of whom have achondroplasia (dwarfism), and who wish to conceive a child who expresses the same phenotype as the parents – that is, they want their child to be achondroplasic. A foetus that is homozygous "normal" (i.e. who will be of average stature) or who is homozygous "affected" (a lethal condition) will be terminated. The couple will only continue the pregnancy if the child is heterozygous for achondroplasia. The authors state: "many geneticists faced with this situation would refuse to participate in this part of the request. A decision that deliberately worsens the clinical status of the offspring would seem to violate the principle of beneficence for the child."[119]

While the authors are consistent in their disapproval for termination considered unnecessary in both cases (mild learning disability versus homozygous "normal"), they seem to place greater value on a child with a learning disability than a child with achondroplasia, an attitude that is potentially devaluing to people with achondroplasia. The suggestion that selecting for achondroplasia would worsen the clinical status of the child is also devaluing. While the issue of selecting for particular characteristics in a foetus is ethically relevant, the question most relevant to this discussion is, what exactly does it mean to have a worse clinical status? Is selecting for achondroplasia, or Down Syndrome, or Deafness worse than selecting for a "normal" foetus? Is it a clinician's place to make this judgment?

In the chapter within the same text, entitled "The Role of Genetics in Medicine," the authors state that "approximately 3% of all pregnancies result in the birth of a child with a significant genetic disease or birth defect that can cause crippling, mental retardation, or early death... the chronic nature of many genetic diseases imposes a heavy medical, financial, and emotional burden on affected patients and their families, as well as society at large."[120] The authors go on to emphasise further the correlation between disability (referred to as "disease") and a burden to society: "it is estimated that at least 10% of adult hospital admissions are as a result of genetic diseases. Thus, the financial and medical burdens of genetic diseases are indeed significant."[121]

This view offers a biased view of disability to genetic counsellors by demonizing genetic difference, and people with genetic differences, as being unequivocally burdensome. No mention is made of the value of people with disabilities, except perhaps in the chapter's introductory quote, a phrase attributed to William Harvey that espouses the clinical utility of the study of "rare forms of disease."[122] That is, people with disabilities present conditions that would otherwise be unavailable for study. In this instance the text seems to be characterizing people with disabilities as basically useless except as subjects for study.

## II. Non-directiveness versus Bias in the Counselling Session

Along with the devaluing ways in which disability is approached in the course materials examined above, also at issue is the fashion in which various authors refer to the necessity of non-directiveness in genetic counselling. For example, in the book *Emery's Elements of Medical Genetics*, in the conclusion of the chapter entitled "Mathematical and Population Genetics," the authors state that "non-directive genetic counselling could lead to a reduction in the number of affected children being born."[123] A process described as being non-directive is in fact correlated with an obviously directed outcome, i.e. a reduction in the number of children with disabilities. If counselling were truly non-directive, would it necessarily lead to a reduction in the number of affected children? The meaning of this sentence would not change substantially if the phrase "non-directive" were removed. The authors go on to speak of the "overall burden of these disorders in terms of human suffering." Again, no mention is made of the worth or value of people with genetic differences except in terms of their scientific utility.

This seemingly contradictory use of terms is also found in the American Society of Human Genetics' definition of "non-directiveness."[124] According to this definition, non-directiveness means:

1. Helping clients understand their options and the present state of medical knowledge, so they can make informed decisions;

2. Helping clients adjust to and cope with their genetic problems;

3. The removal or lessening of patient guilt and anxiety;

4. Helping clients achieve their parenting goals;

5. The prevention of disease or abnormality.

A recent paper[125] analyzes the results of a 1985 survey of genetic counsellors. It found that close to 100 per cent of "683 geneticists in 19 nations" agreed with this definition. These professionals are agreeing with a definition that contains an inherent contradiction between two concepts: non-directiveness and the prevention of disability. If professional practice is directed towards "the prevention of disease or abnormality," how can that practice be considered non-directive?

The text by Mueller and Young, like those by Moore and Persaud, Nora and Fraser, and Gelehrter et al, also uses terms such as "congenital abnormality" and "birth defect" to describe genetic difference, and state that "these terms are perfectly acceptable for the purpose of lumping together all these abnormalities when studying their overall incidence."[126] Also evident is a clear categorization of what is "good" versus what is "bad" concerning the possibility that a foetus is "affected with a genetic anomaly." For example, in a discussion of the calculation of risk in genetic counselling, it is stated that:

> It is also important that genetic counsellors should not be seen exclusively as prophets of doom... the good side of the coin should also be emphasized, particularly if the odds strongly favour a successful outcome. For example a couple faced with a probability of 1 in 25 that their next baby will have a neural tube defect should be reminded that there are 24 chances out of 25 that their next baby will not be affected.[127]

A "successful outcome" is portrayed as one that results in the birth of a child, who is "normal," who does not have a "congenital abnormality" or "birth defect." It is not, for example, an outcome in which a child is born with Trisomy 21 instead of the lethal Trisomy 18, or in which a child is born with an intellectual disability free of potential physical complications such as congenital heart problems. The presence of disability is characterized as *a priori* unfortunate, unsuccessful, a worst-case scenario.

Mueller and Young go on to ask the question of how genetic counselling should be assessed and evaluated. It is stated that the three main outcome measures presently employed within the profession are 1) client recall of information provided by the genetic counsellor or team, 2) impact of information on subsequent reproductive behaviour, and 3) patient satisfaction.

The second outcome measure – impact on subsequent reproductive behaviour – is relevant to this discussion. It is written that the reproductive behaviour of approximately 50 per cent of couples that received genetic counselling was affected in some way. Variables that affected reproductive behaviour are listed as:

- Severity of the disorder
- Desire of the parents to have children
- Availability of prenatal diagnosis and/or treatment.

The outcome measures outlined above are given in response to the perceived ethical problems inherent in evaluating genetic counselling services according to number of births prevented, or heath care costs saved as a result of prevented births. It is said that health professionals in counselling and genetics reject the use of outcome criteria such as the number of abnormal pregnancies terminated and the number of individuals screened

> ...on the grounds that they hint at a eugenics philosophy which is totally unacceptable in a society in which individual choice is paramount. Instead they emphasize the benefits of an educated, informed community with enhanced individual autonomy. The goals of satisfaction as expressed by the users of genetic services and their ability to make informed decisions are seen as much more acceptable than a reduction in the financial and personal burdens caused by genetic disease.[128]

The use of the terms "financial and personal burdens" and "genetic disease" is notable within a supposedly neutral framework. Also relevant to this discussion is the authors' warning of the dangers of "an increasingly cost-conscious society."[129] The responsibility for a family's choice of whether to continue or terminate a pregnancy is placed firmly on the family itself, thereby relieving health professionals of any ethical baggage. The "hint" of eugenics is sidestepped since potential parents are theoretically never counselled to abort a foetus at risk of developing a disability. The authors seem to be saying that it is acceptable (or non-eugenic) for parents to make the decision to terminate a foetus with a genetic difference, as long as a health professional cannot be held responsible for influencing that decision.

In the introduction to the chapter "Prenatal Diagnosis of Genetic Disease," it is stated that "until recently, couples at high risk of having a child with a genetic disorder had to choose between taking the risk or considering other reproductive options such as long-term contraception, sterilization and termination of all pregnancies. Other alternatives included adoption, long-term fostering and artificial insemination by donor sperm."[130]

This phrase provides six different avenues of avoiding the birth of a child with a disability. The only other option – "taking the risk" – is presented as a situation in which parents hope that their child will be "normal." There is no option presented for those who would accept the birth of a child with a disability without fear of suffering and burden, as if that were not a viable option. Near the end of the chapter, it is stated that

> ...so far in this chapter attention has focused on the prenatal diagnosis of abnormalities with the subsequent option of termination of pregnancy. While this is the only option in most situations, there is cautious optimism that prenatal diagnosis will, in time, lead to the possibility of effective treatment in utero.[131]

Again, the discussion centres on avoidance of genetic difference without consideration of the lives of those who actually live with disabilities. Two solutions to the presence of genetic difference in a foetus are presented: termination of the pregnancy, and the prospect of (presently unavailable) pre-conception gene therapy to eliminate the difference.

In another text used in the education of genetic counsellors, population screening is described as "a service seeking a disease, in contrast to the provision of acute medical care, where disease seeks a service. There is a very real danger that genetic screening may lead to the perception that carriers are abnormal and diseased."[132] This is an argument that has much in common with the disability rights critique of prenatal diagnosis.[133] However, the author does not extend this argument to the status conferred by such programs upon people with disabilities. Chapple makes a distinction between genetic carrier status and phenotypic expression of genetic difference. For example, Chapple writes that "genetic screening affects future generations rather than the individual tested since carriers of recessive disorders are not diseased."[134] The author's standpoint is that while carriers are "normal" (and even a benefit to society, since carrier status has certain benefits[135]), those who express the gene are in fact diseased and undesirable.

Chapple goes on to say that "genetic screening services should be available freely for those who want them, but should not be forced upon anyone, and should not be developed at the expense of services for those who suffer from genetic disease."[136] This quote is instructive in two ways: first, the language of the statement is interesting, couched as it is within the terminology of human rights. While advocating freedom of choice, it continues to frame the alternative to testing as a chance that a child will "suffer" from "genetic disease." Second, it illuminates a problematic aspect of programs such as the maternal serum marker screening programs (MSS) in Ontario and Manitoba – that is, the screening test is offered to all pregnant women, which raises the issue of pressure to submit to testing, and the inherent definition of genetic difference as something to be avoided. This subject has also been broached by Mueller and Young, who found that a physician-initiated verbal request to undergo prenatal testing yields a higher acceptance rate than a written invitation. One must question a woman's real autonomy of choice when physicians and the provincial government actively promote testing targeted at reducing the number of babies born with Trisomy 21 and neural tube defects as a necessary part of reproductive health.

Chapple insists on blaming the problems that result from the presence of genetic difference on the conditions themselves, whereas no attention is directed to the social aspects of these problems – prejudice, discrimination, and environmental barriers.

Adding to this, children with disabilities are presented as having only a negative effect on their families. No discussion is presented of the value of these children, nor is the possibility that they have value even considered. For example, the text states that:

> There is no doubt that single gene defects cause a considerable amount of emotional, psychological, social and financial distress to families and the community in which they live. The dangers of assessing screening programs solely on a financial cost-benefit basis have already been stressed, but people with single gene defects need considerable help from health services, social services, voluntary agencies and their families which it is impossible to quantify completely or to price in either financial or social terms.[137]

While Chapple points out the dangers of judging prenatal screening in terms of cost-effectiveness, the author goes on to state that:

> ...parents who love and cherish their affected children always have some regrets at the time they were unable to devote to their other children or with each other, because they had to spend time coping with their child's handicap.[138]

Thus genetic counsellors are again presented with a biased biomedical view that correlates disability singularly with the presence of a genetic condition, and not on the handicapping nature of the social environment. The author's words "spend time coping with their child's handicap" reflects ignorance of the realities of most families. As well, they fail to raise awareness of the exclusionary nature of our society and the resulting need for parents to spend time advocating for services to support their children with disabilities to the same degree as their children without disabilities.

Chapple's description of a "successful" screening program is instructive as to the values underlying this author's point of view. In a discussion of a Thalassemia screening program entitled *Thalassemia – A Success Story*, the author writes of "startling reductions in the number of Thalassemia births,"[139] and provides statistics that show that 50 to 95 per cent of these births were prevented in selected years. The author attributes this "success," among other causes, to "genetic counselling, prenatal diagnosis and high acceptance of termination of pregnancy, even among Catholic parents." It is mentioned that, "only 1% of these children were born as the result of parental choice to continue with an affected pregnancy. Four-fifths of the affected children were born because of parental or professional ignorance of the screening programme instead of informed choice."[140]

## 5.3 Discussion

The work of Susan Wendell provides some insight as to the ways in which the lives of people with disabilities are devalued in the prevailing medical view of disability, and the cultural and societal norms this view reflects and is reflected in.[141] She writes of the cognitive and social authority of health professionals, an authority that informs other professional sectors such as insurance, the legal system, government, employers, and the school system. Wendell cites the danger of morally and ethically biased recommendations being offered by supposedly morally neutral and objective professionals.

These recommendations are powerful because the authority of medicine affects the ways in which people experience their bodies, the ways in which bodies are validated or invalidated, and the types of conditions that become worthy versus unworthy of intervention. In other words, the priorities of medicine and health are reflected strongly in the greater cultural environment of a society. Of course, this argument can be turned around – it may very well be the case that medical philosophies and objectives are informed by and reflective of the culture within which they operate. In either case, the philosophies examined in this review have the same effect – a global, generalized negative view of the lives of people with disabilities.

This philosophy of devaluation also has implications for women's relationships with their own bodies. Wendell writes that the medical experience of the body has become the authoritative version of reality, replacing personal experiences of the body. Hence a third party, i.e. a health professional, mediates one's relationship with one's own body. Wendell is also of the opinion that this authority is compounded for those who have traditionally held less social and cognitive authority, including women, seniors, people with disabilities, and people of ethno-racial minorities.

Landsman puts forward a related concept in the description of a technology-mediated "conceptual separation of the woman from the foetus."[142] As a result of this separation, an adversarial relationship may be created between mother and foetus, wherein the rights of the future child are placed at potential odds with a mother's reproductive choices. Notable here is the increasingly viable and accepted notion that children, particularly children with disabilities, have a right not to have been born (as evidenced by various wrongful birth and wrongful life suits in Canada, the United States, and France). This situation is compounded by a medical philosophy that views disability as inimical to life.

On a more personal level, what does this devaluation mean for people with disabilities? In a discussion of the repercussions of the normal/abnormal split, Linton states that these two categories:

> …affect individuals' most private deliberations about their worth and acceptability, and they determine their social position and societal response to behaviour. The relationship between abnormality and disability accords to the nondisabled the legitimacy and potency denied to disabled people.[143]

The texts examined above may well serve to direct societal responses to people with disabilities. Linton writes of the ways in which discrimination against disability has been woven into cultural norms and meanings of which people may not be consciously aware. Discrimination is also present in "policies and practices that serve to control and marginalize" people with disabilities.[144] While prejudice is no longer as obvious or active as it may once have been, it has by no means disappeared.

Mueller and Young's statement that it is now unacceptable to measure the success of genetic counselling sessions in terms of the number of "abnormal" births prevented must be considered in light of this. Such a measure would provide a clue as to the eugenic origins of prenatal diagnosis and genetic counselling. Instead of couching the discourse of prenatal genetics in terms of a benefit to society (for example, in terms of health care costs saved as a result of the termination of foetuses with genetic differences), services such as prenatal diagnosis and the provision of gene-related information by health professionals is therefore said to be provided for the good of individuals – individual women and families benefit from prenatal services in that they are able, if they so choose, to avoid the "personal burdens" associated with "genetic disease." As Mueller and Young write, "the goals of satisfaction as expressed by the users of genetic services and their ability to make informed decisions are seen as much more acceptable than a reduction in the financial and personal burdens caused by genetic disease."[145] Stated as such within a textbook directed exclusively at members and future members of the medical-genetic community, it serves to sanitize the ethically questionable goal of reducing births of children with genetic differences by using a rationale that provides an appearance of ethical correctness: It's not eugenic if parents "choose" to terminate.

Various studies by Wertz et al. have provided evidence that members of the medical/genetic community are opposed to the birth of children with many types of genetic differences.[146] The employment of language that places responsibility for these births on individuals, and that characterizes genetic professionals as disinterested sources of benign information, does not necessarily represent the reality of the environment surrounding prenatal diagnosis. This environment is one in which disability is equated with disease, and in which the cure or eradication of disease is the overarching goal.

However, the characterization of genetic difference as "disease," "defect," "abnormality," and as something that causes suffering and dependency is one that has been challenged.[147] These kinds of generalizations are based in a dualistic, binary mode of thinking that ignores that the human condition lies along a continuum of difference where there is no "ideal." Thus defining an individual as "healthy" or "unhealthy" based on the arbitrary definition of some traits as being more valuable than others is biased and holds little critical value. It is significant that health and well-being are defined within a hierarchical structure that places a physical ideal of health at the pinnacle. As Wendell writes,

> ...a physical ideal gives us the goal of our efforts to control the body, and the myth that total control is possible deceives us into striving for the ideal... in a culture which loves the idea that the body can be controlled, those who cannot control their bodies are seen (and may see themselves) as failures.[148]

Val Plumwood writes of the way in which Western thought can be defined by this dualism, for example in concepts such as male/female, mind/body, and civilized/primitive. Furthermore, the author claims that these dualisms are based upon a structure of hierarchy – of domination and subordination. In this instance, the healthy and able-bodied constitute the dominant form, while those considered unhealthy or disabled are subordinate.[149]

The argument can also be extended to the medical-genetic concept of a perfect baby with ten fingers and ten toes, of sound mind and body, who will go on to achieve a series of age-appropriate developmental markers. Individuals deviating from this norm are conceived within the profession as worthy of being singled out for examination and possible termination. Moreover, this distinction is provided within an environment that is either dominated by a health professional (i.e. obstetrician, family physician) or one that is purported to be objective and value-neutral (i.e. a genetic counselling session). As Kamarck-Minnich writes, "objectivity and neutrality are not quite the same, but both, attributed to a paradigm, method, theory, or attitude, give it a privileged, superior status in the dominant culture."[150]

The materials outlined in this review present ideals and viewpoints that are neither neutral nor objective, but which are handed down from supposedly objective givers (i.e. physicians) to receivers (i.e. patients) of knowledge. This has two interconnected implications: first, the presence and dissemination of a medically-sanctioned view of genetic difference that is negative and devaluing of people with disabilities, and second, the provision of prenatal care to women that is imbued with pressure to undergo "normal," "beneficial," and therefore "necessary" prenatal screening and/or diagnosis aimed at preventing disability, disease, suffering, and financial and emotional burden.

# NOTES

[81] Society of Obstetricians and Gynaecologists of Canada (1998).

[82] Wertz et al. (2001), p. 12.

[83] Ibid.

[84] Farrant (1985), p. 664.

[85] Marteau (1990).

[86] For elaboration, see Turnbull (2000); Parens & Asch (1999); Terrell-White (1999); Vehmas (1999); Gallagher (2001).

[87] Cocks (2000), p. 515.

[88] Linton (1998), p. 25.

[89] Hughes (2000); Oliver (1990).

[90] Swain & French (2000), p. 569.

[91] HRDC (1998), p. 1.

[92] See, for example, Korf (1996).

[93] For example, Mueller & Young (1998); Nora & Fraser (1994).

[94] For example, Bartels et al. (1993); Marteau & Richards (1995).

[95] Such as Edelson (2000); Featherstone (1981).

[96] Moore & Persaud (1998).

[97] Footnote, p. 172.

[98] Marteau & Slack (1992).

[99] Ibid., p. 693.

[100] Marteau & Richards (1995).

[101] Ibid., p. xiv.

[102] Ibid., p. xv.

[103] Nora & Fraser (1994).

[104] Ibid., p. vii.

[105] Ibid., p. 291.

[106] Ibid., p. 292.

[107] Ibid., p. 293.

[108] Nora & Fraser (1994), p. 293.

[109] Ibid., p. 293.

[110] See Hughes (2000); Linton (1998); Oliver (1996).

[111] Hubbard (1997), p. 187.

[112] Nora & Fraser (1994), p. 299.

[113] Ibid., p. 301.

[114] Ibid., p. 301.

[115] Ibid., p. 291.

[116] Ibid., p. 303.

[117] Gelehrter et al. (1998).

[118] Gelehrter et al. (1998), p. 332.

[119] Ibid., p. 333.

[120] Ibid., p. 2.

[121] Ibid.

[122] Ibid., p. 1.

[123] Mueller & Young (1998), p. 7.

[124] Fraser (1974).

[125] Wertz & Fletcher (2001).

[126] Mueller & Young (1998), p. 242.

[127] Ibid.

[128] Ibid., p. 245.

[129] Ibid., p. 244.

[130] Ibid., p. 303 .

[131] Ibid., p. 320.

[132] Chapple (1992), p. 581.

[133] See Parens & Asch (1999).

[134] Ibid.

[135] For example, carriers of certain types of Sickle Cell Anaemia have increased immunity to Malaria.

[136] Chapple (1992), p. 583.

[137] Ibid.

[138] Ibid.

[139] Ibid., p. 587.

[140] Ibid.

[141] Wendell (1996).

[142] Landsman (1998), p. 79.

[143] Linton (1998), p. 34.

[144] Ibid., p. 34-35.

[145] Mueller & Young (1998), p. 245.

[146] Wertz (1998); Wertz et al. (2001); Wertz & Fletcher (2001).

[147] See, for example, Oliver (1989); Roeher Institute (2001); Reindal (1999); Williams (1996).

[148] Wendell (1997), pp. 268-269.

[149] Plumwood (1997).

[150] Kamarck-Minnich (1990), p. 86.

# 6

# GENETICS, DISABILITY, AND LAW

## 6.1 Introduction

The purpose of this section is to canvas the Canadian legal regime in which the practice of genetic counselling falls. A review of the legislative history of reproductive and genetic technologies, and of existing policy and professional guidelines in many ways sets the parameters for discussion. It addresses the issues surrounding the legal obligations of health care providers (broadly defined) in the context of genetics and assisted reproduction – abortion, prenatal liability, wrongful birth and wrongful life jurisprudence is presented. Since there has been little Canadian litigation specifically arising from prenatal testing and genetic counselling, the piece points to potentially relevant jurisprudence. Commentary from secondary literature is also included to highlight legal issues of particular relevance to genetic counselling, to consider ethical principles, and to illustrate areas of academic and practical concern. Finally, the existing jurisprudence in the area of "disability" is reviewed. At present, notions of "disability," "risk" and "harm" within law are complex, and often conflicted. This section will demonstrate that depending on the context in which disability is in question, different legal understandings have been applied. Guidelines for genetic counselling must take these complexities into account.

## 6.2 Review of Legislation, Policy, and Guidelines

### 6.2.1 Legislation – The (Potential) Regulation of Reproductive and Genetic Technologies

Currently, there is no specific legislation in effect relating to the use of genetic information in Canada, nor are there statutory restrictions on the practice of genetic counselling.[151] "[A]lthough the training standards for Ph.D. medical geneticists and for individuals with Masters degrees in genetic counselling are undeniably high, no licensing procedure exists thereby leaving the legal status of these practitioners in question."[152] As a result, for genetic counselling, there is no formal standard of competence through which to protect the public. In 1993, the Royal Commission on New Reproductive Technologies noted,

> ... [a]s with variations in referral practices, variations in the way prenatal screening tests are provided reflects the absence of established standards for those practices of GPs and obstetricians in this area, as well as the lack of monitoring of these practices... There are, moreover, no qualifying exams for GPs or obstetricians who wish to provide PND [prenatal diagnosis] services.[153]

Researchers documented that as of 1995, "...[a] great deal of the guidelines and protocols followed by the various [genetic] centres had not been systematically documented or approved."[154]

There is no social consensus in Canada regarding which uses of reproductive and genetic technologies are acceptable. Federal regulation of reproductive and genetic technologies has the potential to set limits for use and research, and to enforce moral and ethical standards within those limits. Regulation has the potential to ensure that the needs of (often vulnerable) individuals as consumers of "health" related services are balanced against, or indeed protected against, those of the commercial/private sector. With respect to genetic counselling, regulation could mandate clearly articulated standards for counselling upon and after use of reproductive and genetic technologies to ensure that those with disabilities do not face devaluation, discrimination, or limited opportunities to experience life.[155] Legislation pertaining to reproductive and genetic technologies could "ensure respect for the fundamental values of Canadian society, protect the public against risks to health and safety, and provide clear principles of law according to which potential disputes could be resolved."[156] This does not assume, however, that the creation of such legislation would be a simple task. The present lack of federal regulatory activity has resulted in situations where reproductive and genetic technologies are governed primarily by the economic marketplace. This creates situations of unequal access, unmonitored development and use, and legal uncertainty for patients/consumers, physicians, researchers, and developers.[157]

In 1995, a voluntary interim moratorium on reproductive and genetic policies prohibited a range of practices.[158] The document, *New Reproductive and Genetic Technologies: Setting Boundaries, Enhancing Health* that set out the proposed elements of a regulatory framework (prohibitions and regulations) was released for public discussion. In June 1996, the *Human Reproductive and Genetic Technologies Act* (Bill C-47) was introduced. The preamble of Bill C-47 began with an expression of grave concern about, "the *significant threat* to human dignity, the *risks* to human health and safety, both known and unknown, and other serious social and ethical issues posed by certain reproductive and genetic technologies."[159] As a result, the Bill attempted to codify 13 prohibitions. The Bill reached second reading in November of 1996, when it was approved by the Sub-Committee on Health. Final reading of the Bill did not occur prior to the dissolution of Parliament with the call of the 1997 federal election.

The proposed legislation was subject to much scrutiny by a wide range of stakeholders. The Canadian Medical Association (CMA), for example, criticized the bill on the basis that it lacked a comprehensive regulatory framework for the new technologies. This framework, according to the CMA should build upon Canada's already "robust regulatory infrastructure."[160] It states,

> ... [t]he optimal way to regulate the practice of medicine in Canada, including the new reproductive and genetic technologies, is through the existing system of provincial self-regulatory bodies and profession-generated standards of practice and practice guidelines. This follows from the principle of subsidiarity and is consistent with the Canadian way of regulating medical practice. In Canada, most areas of medical practice and scientific research are regulated quite satisfactorily apart from legislation; for example, by professional codes of ethics, by the provincial/territorial medical licensing authorities and similar licensing bodies for other professions, and by research ethics guidelines and committees.[161]

Several of the elements of a regulatory framework, as outlined by the CMA, pertain directly to the issue of genetic counselling. These include: the development and monitoring of national standards for training and certifying physicians (presently "appropriately done" by bodies such as the Royal College of Physicians and Surgeons of Canada and the College of Family Physicians of Canada); the licensing and monitoring of individual physicians (currently the responsibility of the provincial and territorial medical licensing authorities); the development of guidelines for medical procedures (normally done by the medical specialty societies, such as the Society of Obstetricians and Gynaecologists of Canada); the accreditation of facilities where reproductive technologies are offered (currently done by the Canadian Council on Health Services Accreditation, a multidisciplinary organization).[162] Regulatory bodies will be examined in the following section.

On May 3, 2001, then Health Minister Allan Rock introduced draft legislation, the *Assisted Human Reproduction Act* (AHRA), pertaining to reproductive and genetic technologies. Again, this draft legislation is intended to prohibit practices such as human cloning, creating embryos for research and commercial surrogate motherhood. Building on the prohibitions contained in Bill C-47, the draft legislation includes both *prohibited activities*, which carry criminal sanctions, and *controlled activities*, which would be regulated through the issuance of licenses. The draft legislation also proposes a regulatory scheme that would allow research on existing embryos and combining human cells with animals within guidelines. The establishment of a national regulatory body to set standards, issue licenses and maintain information registries was also proposed.

The draft AHRA was not introduced directly into the House of Commons. Instead, it was sent to the Parliamentary Standing Committee on Health for its review.[163] The draft legislation contains a preamble setting out a number of guiding principles and considerations that inform the *Act*, which serve to guide lawmakers in interpreting and implementing the legislation. Included in this list are: recognition of the need to "protect and promote the best interests of children," recognition that "women more than men are directly and significantly affected by" the application of reproductive technologies, a commitment to the principle of "free and informed consent" in the use of reproductive technologies, a recognition of the "health and ethical concerns inherent in the trade in the reproductive capacities of women and men", and in their exploitation and that of children for commercial ends, and a recognition of the "importance of preserving and protecting human individuality and the integrity of the human genome."[164] The draft legislation places reproductive and genetic technologies in a more favourable light than did Bill C-47 by focusing on the potential *benefits* rather than the potential *risks* of such technologies.[165]

The draft AHRA regulates both prohibited and controlled activities. Prohibited activities under the draft legislation include: human cloning by embryo separation and somatic cell nuclear transfer; germ-line genetic alteration; the development of an embryo outside a woman's body beyond the accepted 14-day limit; the creation of embyros solely for research; creating an embryo from another embryo or foetus; transplanting reproductive material from animals into humans; the use of human reproductive material previously transplanted into an animal; the sale and purchase of human embryos; sex selection (except for reasons relating to the health of the resulting human being); the purchase, barter or exchange of sperm or ova; and, commercial surrogacy arrangements. These prohibitions are similar to those in Bill C-47. In addition, the draft AHRA contains specific provisions prohibiting the counselling or inducement of a minor to become a surrogate.[166] The prohibition against human cloning and provisions related to minors are the only new items to be added to this list.[167]

The controlled activities listed in the AHRA are permitted only if carried out under license and in accordance with the regulations to the draft AHRA. Controlled activities include: altering, manipulating or treating human reproductive material for the purpose of creating an embryo or facilitating human reproduction; using an *in vitro* embryo, or part of one for research or the prevention, diagnosis or treatment of a disease, injury or disability; exporting, storing or handling gametes or embryos for reproductive or research purposes; the creation of a chimera; combining any part of the human genome with that of an animal species; and, reimbursing the donor of gametes, *in vitro* embryos, human cells or genes for an expense incurred in the course of donating, or reimbursing a surrogate mother for her expenses incurred in relation to her pregnancy.[168] The draft AHRA empowers the Minister of Health with broad discretion to regulate the proposed controlled activities;[169] however, these regulations are not specified in the legislation. The proposed licensing regime does not distinguish among clinical reproductive uses and research uses of reproductive materials, but rather includes them in the same regulatory framework. While Health Canada's promotional literature suggested that the regulations will set out counselling requirements for donors and recipients[170], they are not specifically referred to in section 40(1) of the ADHR, which empowers the Governor in Council to make regulations to implement the *Act*.

The draft legislation recognizes the significance of health information, both as a privacy issue and as an important societal resource for the promotion of health. To this end, the draft AHRA mandates the maintenance of a personal health information registry[171] and a public health registry.[172] The draft AHRA seeks to restrict personal nominative information and endeavours as far as possible to keep the use of such information at the discretion of donors and persons conceived through reproductive technologies.[173] It also mandates the collection of health information for purposes of public health and makes that information available for public health purposes and accessible to the descendants of donors.[174] The draft legislation further mandates access to the information used to evaluate and adjudicate applications for a license.[175] The exact nature of the information to be disclosed for the purposes of public health is subject to regulations, the details of which are not specified in the draft legislation.

The policy framework that accompanies the draft legislation contains "guiding principles," including: individual choice; balancing individual and collective interests; equality of men and women; protection of the vulnerable; appropriate use of technologies for treatment; non-commercialization of reproduction and reproductive materials; accountability; and, integrity. In 1999, Health Canada stated, "[C]onsideration is being given to the range of options that exist for incorporating ethical review into the policy development and regulatory processes surrounding reproductive and genetic technologies."[176] The Parliamentary Standing Committee, in its response to the draft legislation, attempted to give (several of) these principles additional legal "teeth."

In December 2001, the Parliamentary Standing Committee on Health delivered a report, entitled, "Assisted Human Reproduction: Building Families."[177] The report is the Committee's response to Rock's draft legislation. It attempts to balance the well-being of children conceived of assisted reproduction and the health and safety of adults who utilize the procedures, with the potential benefits of research in this field. The Committee situates the former as its priority, and reinforces the need for national legislation on assisted human reproduction and related research. To strengthen the principles contained within the preamble of the draft AHRA, the Committee recommended the removal of the preamble and addition of a statutory declaration *within* the body of the (potential) legislation. By entrenching the guiding principles within the *Act*, the declaration would not only aid in the interpretation of the purpose and objectives of the *Act*, but would give the principles "greater legal significance and effect."[178]

The principles contained within Recommendation 3 of the report aim to enshrine respect for human individuality, dignity and integrity, and declare that the fundamental condition of the use of assisted human reproduction is free and informed choice.[179] It prioritizes the health and well-being of children born of assisted reproduction, and recognizes that women are more significantly affected by the application of the technologies.[180] The recommendation also makes direct reference to people with disabilities. It states that "persons with disabilities can lead full and satisfying lives and enrich the lives of those around them."[181] In so doing, it also asserts the importance of the "protection" of the "integrity" of the human genome.[182]

While the declaratory principles attempt to recognize and value the lives of people with disabilities, they exist in some opposition to the recommendation of the Committee regarding sex selection. The draft legislation addressed the prohibition of such procedures as gamete manipulation and modification of fertilization techniques that would increase the probability of obtaining an embryo of one sex or the other.[183] The Committee, however, desired the additional prohibition of sex selection through genetic pre-implantation diagnosis of embryos. While this addition is in keeping with the principles, Recommendation 9 of the report specifically prohibits all sex selection with the noted *exception* of "disorders" linked to the sex chromosomes.[184] According to the Committee, these exceptions are to be permitted for reasons of "health." The recommendation does not value the lives of those with "disabilities" linked to their sex chromosomes, as it effectively deems people with such "disorders" to be "unhealthy;" their lives potentially not worth living. While the Committee recognized that assisted reproduction "impinges on society's sense of the uniqueness, worthiness, and wholeness associated with being human," the ethical implications of permitting sex selection on these bases do not appear to be of grave concern. Thus, normatively, the Committee has made a recommendation which permits, and arguably promotes, the perception of those with sex-related "disorders" as less whole, less worthy, and less human.

At the time of publication, there was not yet any federal legislation arising from the Committee Report and public scrutiny of the draft legislation.

### 6.2.2 Policy/Guidelines

In the absence of legislation in which ethical principles and standards of care are codified, health care practitioners look to policy and professional guidelines for guidance as to how we can, and ought to, use reproductive and genetic technologies. Policy related to genetic counselling documents issues related to providing "unbiased" information to those who use the technologies to communicate about the (potential) "risks" and "harm" related to genetic testing. Similarly, by attempting to ensure that genetic counselling is "non-directive" and as objective and value neutral as possible, professional guidelines attempt to ensure that the actions of genetic practitioners fall within a (legally) acceptable range of behaviour(s). As such, they focus predominantly on issues of informed consent and confidentiality of information.

In its 1993 policy recommendations, The Canadian Royal Commission on New Reproductive Technologies (RCNRT) stated that the aims of the genetic counsellor are to:

1. Provide estimates, in understandable form, of the probability of having an affected child and on the risks and benefits of contemplated procedures;

2. Provide information about the nature, burden, and possible variability of the disorder and about what treatments and supports are currently possible for a child born with a particular disorder;

3. Try to allay anxiety based on misperceptions and help the woman or couple to deal with that which is well founded;

4. Try to appreciate the couple's perception of risk and burden and where they fit on the spectrum of views regarding abortion, the disabled, quality of life, and attitudes toward life (for example, are they optimistic, realistic, fatalistic?) Every family is different and, recognizing this, counsellors must try not to project their personal views in the situation;

5. Try to help the couple without being directive, to reach a decision best suited to their own situation, by pointing out pros and cons and acting as a sounding board and resource person. The counsellor wants to empathize with the couple or individual, yet must remain objective to be effective. It is the tenet of genetics counsellors not to be directive, fully recognizing that this is not easy.[185]

The RCNRT policy expressly documented the need for genetic counsellors to provide guidance in accordance with each individual's own values, experiences, and situations. It stressed the essential requirement that counselling be as objective as possible.

> Some representatives of people with disabilities worried that doctors are providing patients with stereotyped and inaccurate information about living with disability. They claim that some doctors, particularly, in the referring network, are telling parents that a child with cystic fibrosis or Down syndrome leads a painful or worthless existence. Many parents feel it would be selfish to abort an affected foetus solely on grounds of hardship to themselves; as a result, they believe, there may be a tendency to exaggerate the extent to which a disorder or disability causes suffering to the child... Giving parents biased information would violate the principle of autonomy. It is therefore essential that information be as objective and accurate as possible.[186]

The Royal Commission further stated that

> ... [p]eople receiving counselling should be fully informed therefore, not only about the risks and the disorders involved, and about what the disorder means in terms of day-to-day functioning for the affected individual and lifelong consequences for themselves, but also about the social pressures they may experience, so that they have an opportunity to consider how such pressures might affect them.[187]

The RCNRT signalled the need for the means to communicate with genetic practitioners about what it "means" to live with a disability in as much of a neutral, unbiased manner as is humanly possible. Both stress the importance of non-directive genetic counselling to fulfill this objective. Reproductive and genetic technologies produce important information about our bodies. The uses to which this information may be put are vast, and are highly controversial. Many questions arise as to who "controls," and who can access genetic information. Lack of access to, or lack of understanding of, genetic information may lead to physical and emotional "harm."

> Out of counselling and consent flows a power to exert rights over or, at least to some degree, "control" one's genetic material and information. Arguably, much of this power is exerted in the informed consent process whereby the patient gives his or her consent to the test and to the subsequent use of the resulting information and material (whether for research or otherwise). This notion of control takes on a heightened importance in the field of genetics due to the potential financial gain associated with genetic technology and to the development of various types of DNA banking facilities.[188]

As this quotation demonstrates, genetic medicine is not only a means to ensure the "health" of our population; it is also a producer of a wide range of highly profitable economic commodities within an emerging "health care market." The protection of "health" and the pursuit of profit are not necessarily complimentary goals – particularly when/if people with disabilities are considered "unhealthy." Reproductive and genetic technologies give us the ability to define and alter our perceptions of what/whom is "normal," and "healthy."[189]

In addition to the RCNRT, Health Canada has also identified areas of concern with respect to genetic counselling and our perceptions of disability. In March 2000, Health Canada established the Working Group on Public and Professional Educational Requirements Related to Genetic Testing of Late Onset Disease. One of the stated policy goals of the group is to "[i]dentify the educational/learning needs regarding genetic testing of late onset diseases among health professionals and the public."[190] Within the document, "Genetic Testing for Late Onset Diseases: In-depth Thematic Analysis of Policy and Jurisdictional Issues," the relationship between public policy and genetic technologies was studied in the contexts of the difficult nature of evaluating genetic technologies, the potential of a "balanced" approach to developing genetic policies and regulations, and in terms of the available approaches for regulatory decision making.[191] Since the focus of the article is late onset disease, issues related to genetic counselling in the context of prenatal testing are not addressed. However, when assessing the potential risks associated with genetic testing (for late onset disease), it is made clear that at present, the risks outweigh the benefits. This is due to several factors including: the present lack of treatment available to those who test positive; the potentially discriminatory results of undergoing genetic testing when/if the genetic information becomes available to insurers or employers; and, concerns about the impacts of genetic testing on family relationships and individual self image.[192] The report does specifically refer to the documented need for genetic counselling, however, it re-articulates that the combination of limited (state) resources devoted to genetic testing and the private (i.e. over the Internet) availability of genetic testing renders genetic counselling inadequate.

Four key ethical issues were studied within the report: genetic discrimination (including issues of privacy, confidentiality, and the protection of vulnerable populations), the issue of patent law in relation to genetic material, the marketing and allocation of genetic resources, and the issue of scientific freedom (including the issues of individual rights and autonomy).[193] In its treatment of genetic discrimination, the report focuses on the potential threats to insurability and employability arising from information gained through genetic tests. The second concern raised in the context of discrimination is that of privacy – the potential "genetic" intrusions into individual treatment, reproductive, and lifestyle choices.[194] The report argues that regulation of data collection and storage is necessary to protect vulnerable populations. Although, at present, the group is concerned with issues relating to insurance and employment, the report does mention the potential of "genetic essentialism" to lead to racial and ability-based discrimination, which "can also lead to an uncritical acceptance of what is 'normal' and 'acceptable' about being human."[195]

In addition to policy, professional guidelines exist which delineate the responsibilities of health care practitioners who utilise genetic medicine, and provide genetic counselling. Health care professionals are regulated to protect the public – from the perils of incompetent practices, and from the "imperfect" market conditions in which health care exists.[196] This market is characterized by "the risk and uncertainty associated with contracting illness (it is generally not possible to plan one's consumption of health care services), externalities [such as the lack of knowledge of one's health status, and of available services – each having the potential to affect the health of others]; and, the asymmetrical distribution of information about health care between providers and consumers (also called "information failure")."[197] However, professional regulation is not a perfect control mechanism. Government intervention into the health care market in the form of occupational regulation confers a great deal of power upon the professions themselves.[198]

Regulatory control of a profession can take the form of proactive input regulation (i.e. restricting who can provide a service) or output regulation (i.e. civil liability or professional monitoring/discipline).[199] Certification and licensing, characteristic of input regulation, are the proposed methods of regulation of health professionals who work with assisted reproductive and genetic technologies. At present, however, with the lack of formal regulations pertaining to the technologies themselves, health professionals in this field are largely self-regulated. Self-regulatory regimes are

> ...created by provincial statutes which delegate regulatory functions to a profession. This delegation of authority is generally made to a body created by the statute and charged specifically with the protection of the public interest, which may be called a "college," an "association," a "council," or a "board." The statute delegating regulatory functions and outlining the associated responsibilities can be a stand alone statute (i.e. specific to the particular profession), or it can be an umbrella statute which establishes self-regulatory bodies for a number of health disciplines under a common framework.[200]

The governing bodies of self-regulating professions are responsible for controlling who enters the profession, and monitoring their conduct and competence after admission to the profession.[201] While self-regulatory regimes are often a less costly and more efficient method than government administration, they are often characterized by self-interest on the part of the health profession. "One of the main concerns associated with self-government is that the interests of the profession might conflict with the interests of the public."[202] One such "public interest" – particularly in the context of genetic testing – is the protection and valuation of persons with disabilities.

An investigation of the guidelines of several of Canada's predominant professional bodies practising genetic medicine – the Royal College of Physicians and Surgeons, the Canadian College of Medical Geneticists, the Canadian Association of Genetic Counsellors and the Society of Obstetricians and Gynaecologists – illustrates the current standards and norms applied by genetic practitioners. Since these are self-regulating professions, these guidelines are distinct from legislation and the common law. "Canadian [and American] professional accreditation standards for medical genetics/genetic counselling provide a framework of appropriate skills, attitudes, and clinical objectives, but it remains unclear how such standards are to be interpreted in light of an appropriate legal standard of care."[203] The professional accreditation standards for medical genetics/genetic counselling will be canvassed, as they provide a framework of what these bodies consider "appropriate" skills, attitudes and clinical objectives.

> Genetic information is conveyed to patients by a variety of professionals including nurses, obstetricians, Ph.D. geneticists, medical geneticists, general practitioners and specifically trained genetic counsellors. Each type of practitioner approaches the counselling process differently, and each may be subject to a different set of guidelines, professional obligations, legal duties and ethical mandates.[204]

Generally, professional accreditation standards for medical genetics highlight the importance of communication – they are designed to provide guidance on the issue of the duty of care owed by the physician. The Royal College of Physicians and Surgeons of Canada (RCPS) requires physicians to have the ability

> ...to communicate at a level appropriate to the consultant or the referring physician, information concerning the medical implications and prognosis, the risks that apply, the options available; to help the individual/family choose an appropriate course of action for themselves; and, be able to advise them regarding support agencies.[205]

As such, a medical geneticist must be able to "describe and discuss" genetic screening and testing in relation to federal and provincial laws related to genetic diseases, reproductive options and technology, and those relating to confidentiality, autonomy, disclosure, privacy, and issues of competence.[206] As this attempt to canvass the legal regime in which genetic counselling falls will demonstrate, however, this is not a simple task (particularly for those who do not have legal training). As part of a medical geneticist's clinical knowledge, the RCPS also mandates that a practitioner must be able to make the distinction between genetic testing for the diagnosis of disease, and predictive testing to assess risk for predisposition to monogenetic or complex genetic diseases (as well as their applications and limitations).[207] The specific requirements of the geneticist as "communicator" include the ability to

> ...[p]rovide genetic counselling: display empathy and compassion, especially in delivering bad news, remaining objective and impartial, remaining appropriately non-directive, but being prepared to advise in certain situations and to provide psychological support either personally or through referral, employing active listening skills, delivering information to the patient and family in a manner that is understandable, encouraging discussion, and promoting patient and family participation in decision making.[208]

Also required within the communication process is the ability of the practitioner to assess values, beliefs, concerns, and expectations. The practitioner must demonstrate the ability to

> …[g]ather information not only about the disease but also about the patient's beliefs, concerns and expectations about the disorder, while considering the influence of factors such as the patient's age, gender, ethnic, cultural, and socioeconomic background, and spiritual values; recognize one's own biases, including ethno-cultural differences, and their impact on communication and patient care; and, understand how cultural background, age, gender, socioeconomic background and spiritual values influence communication.[209]

The RCPS situates these requirements within a particular understanding of "medical genetics." It understands this branch of medicine as one, "concerned with the effect of genetic variation on human development and health and also with the study, diagnosis, management, and prevention of disease in individuals, families, and communities."[210]

The Canadian College of Medical Geneticists requires training in "psychological dynamics, stress management and crisis intervention... and… how to recognize the psychological implications of a genetic problem in the family, with an ability to provide psychological support either personally or through referral."[211] Similarly, the Canadian Association of Genetic Counsellors (CAGC) has developed a "Scope of Practice," which serves as the basis for the development of a certification process. The responsibilities of the genetic counsellor include practical and ethical dimensions. Genetic counsellors should be able to apply the ethical principles of autonomy, non-maleficence, beneficence, justice and paternalism to genetic counselling cases.[212] Counselling, according to the guidelines, should be client-centred and non-directive. When obtaining medical history from a patient, genetic counsellors are to obtain "appropriate" medical, social and psychological histories.[213] "Appropriate" is not further defined. As part of the duties of the genetic counsellor, the guidelines outline that the stages of grief and "unhealthy" grief should be understood. In addition, "bereavement issues" arising in the following contexts should be able to be "described" by the counsellor – pregnancy termination, miscarriage or stillbirth, birth of a handicapped child, or diagnosis of a genetic condition in a child.[214] The CAGC also mandates that genetic counsellors have an "awareness" of multicultural society. This is to be accomplished by

> …demonstrating an appreciation of diverse cultures, and an understanding of the difference between your beliefs and those of your client; identifying institutional barriers that prevent minorities from fully accessing genetic counselling services; having an awareness of culturally specific methods of helping; and, understanding how the value assumptions on which the theories of genetic counselling are based may interact with the values of different cultural groups.[215]

According to the CAGC guidelines, genetic counsellors should also be "familiar" with legal issues stemming from genetic counselling including (but not limited to), discrimination, competence, standard of care (practice), laws governing abortion, and the components required for informed consent.[216] In the specific context of prenatal testing/diagnosis, counsellors should have an understanding of the indications, advantages, limitations, risks and accuracy of prenatal procedures."[217] Parental age is a specific risk factor requiring understanding according to the guidelines. Specifically related to disability, genetic counsellors should be able to

> …demonstrate an understanding of risk factors as they relate to the risk to a fetus of having a birth defect, syndrome, or adverse outcome; demonstrate an understanding of testing

> done to determine if a pregnancy is at risk for mental retardation, birth defects, premature delivery, or infection. Know when such testing is appropriate and be able to interpret test results; [and] demonstrate an understanding of the technological and psycho-social issues related to recurrent miscarriages, in vitro fertilization, and ICSI.[218]

The Society of Obstetricians and Gynaecologists of Canada (SOGC) has also developed practical guidelines for those professionals offering prenatal tests, and for genetic centres providing counselling and specialized prenatal screening and diagnostic testing.[219] The guidelines mandate access to qualified professionals, knowledgeable in the field of genetics, and contain specific recommendations pertaining to the process of genetic counselling. According to the SOGC, each patient should have an assessment of family history and genetic counselling prior to undergoing prenatal diagnosis.[220] Counselling should

> ...be given in a non-directive manner and should include a discussion of the risks and benefits of the various types of prenatal diagnosis procedures. Patients should be reminded that normal test results do not guarantee a normal foetus.[221]

The guidelines, however, do not articulate the nature of "non-directive" counselling, nor do they make explicit the standard of "normalcy" to which they refer (or to which liability might attach).

In its 1998 policy statement, the SOGC adds some clarity to the issue of non-directive counselling. "Counselling of the woman and her partner should be non-directive and comprehensive, in a language they understand. Women considering prenatal testing should be made aware that agreement to terminate a pregnancy if an abnormality is found is not a precondition or requirement prior to counselling or testing."[222] According to the SOGC, genetic counselling should include a discussion of

> ...available methods of prenatal diagnosis; risks and benefits of various techniques; timing of prenatal procedures; the condition(s) for which testing is to be considered and current management of relevant genetic disorders; accuracy of prenatal test results (including the implications of false-positive and false-negative results where appropriate).[223]

The option of abortion should be discussed within counselling, and may be accompanied by written information from a local centre. The policy statement outlines three conditions in which referral to a genetic or prenatal centre is appropriate: "...when invasive testing is indicated, when there is a significant family history or if the woman and/or partner request additional information."[224] At this time, care must be taken to communicate complete and accurate information to genetic centres or laboratories performing the genetic testing. "Patient-specific factors," such as age must be conveyed as they can influence the interpretation of some screening tests (i.e. maternal serum screening risk estimation).

Prenatal diagnosis counselling, according to the SOGC, has an important informational content. "Appropriate" written information and other educational aids can be made available for couples attending genetic clinics – although "appropriate" is not defined, nor is it clear who makes these decisions of "appropriateness." The information provided by the genetic centre should be regularly reviewed and updated – again, no clarity as to the meanings intended by these terms was provided. The SOGC provides that prospective parent(s) should be given "sufficient time" to weigh the options of alternate screening or diagnostic procedures. "Where practical, counselling appointments should be scheduled at least one day prior to invasive procedures so that couples have time to think about their options and do not feel pressured into having genetic testing."[225] Telephone discussions are presented as an important method of communication for women geographically distant from a centre.

In the situation where testing is offered to a woman before adequate research into the accuracy and/or safety of the test, "great care must be taken to ensure that she understands the uncertainty and the experimental nature of the proposed investigations."[226] The SOCG states that a "detailed explanation outlining the research nature of the procedure *should* be given."[227] Hospital ethics committees or provincial advisory committees should review new testing procedures, according to the SOCG, to ensure consistency with any provincial or national guidelines. In the absence of such guidelines, however, the counselling process becomes paramount. "[T]hese procedures may be offered in specific situations only after detailed discussion with the woman about the possible advantages and disadvantages of using a testing procedure that lacks sufficient evaluation."[228]

Similarly, guidelines for conduct by genetic researchers include discussions of genetic counselling to ensure that research protocols make provisions for access to genetic counselling for the subjects.[229] The Natural Sciences and Engineering Research Council of Canada (NSERC) policy statement situates genetic counsellors as having two main roles: to educate about particular conditions, and to present options or scenarios in a "non-directive" manner.[230] NSERC recognizes the potentially competing interests at stake as a result of the development and use of assisted reproductive and genetic technologies.

> Given the present inability to know the limits or effects of such research, or the context in which genetic information is interpreted and used, caution should be exercised. These rapid changes and the potential financial gain from marketing the technologies drive the need to be sensitive to ethical issues in genetic research. The potential ability to identify all human genes and their mutations has profound social implications. Misunderstanding or misuse of the results of genetic testing has the potential to interfere with an individual's self-identity and sense of self-worth, and to stigmatize the entire group to which that individual belongs.[231]

As such, the policy statement attempts to illustrate its "eugenic concerns." It states, "[t]he aim of genetic research should be to advance knowledge or to alleviate disease, not to 'improve' or 'enhance' a population by cosmetic manipulation… care should be taken to avoid isolating specific populations so that the group [n]either feels stigmatized by the genetic disorder [n]or targeted for 'improvement.'"[232] It states:

> The rights and freedoms attached to personal relationships, reproduction, and the support of those with handicapping conditions should also be maintained. The freedom of couples who are at risk to plan and carry potentially affected pregnancies, and the support of children and adults with handicapping conditions, should not be compromised.[233]

Those who carry out genetic counselling and research studies, "*shall* seek free and informed consent from the individual and report results to that individual if the individual so desires."[234] The NSERC policy attempts to make researchers aware of the complexities of obtaining informed consent in the genetic context by bringing to light tensions between individuals and families. It cautions researchers that individuals may be coerced by their family members to participate, that it may be difficult to obtain the consent of all affected family members, and to be aware that tensions may be created within families by the lack of/desire of certain members to participate. Privacy issues are also addressed within the policy statement. Researchers have a responsibility to "ensure that the results of genetic testing and genetic counselling records are protected from access by third parties, unless free and informed consent is given by the subject. Family information in databanks shall be coded so as to remove the possibility of identification of subjects within the bank itself."[235] Harms to a participant and her/his family are also an important issue stemming from genetic research. NSERC states that researchers and genetic counsellors *shall* "reveal potential harms to the Research Ethics Boards (REB) and outline how such harms will be dealt with as part of the research project."[236] Moral, physical, psychological, and social harms are to be documented. The risk of harm to and stigmatization of children is addressed within the policy. It states that "genetic research involving children should not be done unless an effective intervention is available and the information to be gained outweighs the risk of harm."[237] Harm, within this policy, is balanced against the ability to treat a genetic "condition" once identified. The policy suggests that testing should not occur where preventative measures do not yet exist, but may be appropriate where the "knowledge [obtained] affects treatment options."[238]

The NSERC policy states that, "genetics researchers and the REB shall ensure that the research protocol makes provision for access to genetic counselling for the subjects, where appropriate."[239] Genetic counselling, according to the NSERC policy, augments or supplements informed consent, which is required. The policy states: "[t]he complexity of genetic information and its social implications usually requires that free and informed consent be *supplemented* with genetic counselling."[240] The potential benefits, harms, and limitations of each study must be communicated to the participant at two stages in the research process – before the individual gives free and informed consent, and after results are available. Further, when the clinical implications of genetic research remain unknown, the policy creates a duty for the researcher to advise the research subject and the REB about "the potential meaning of the anticipated results to the subject, and how counselling will be handled."[241] Within the policy statement, there remain questions about the duration of follow-up counselling and when exactly the researcher's obligation ends. Although prudent follow-up and referral to a physician/geneticist by the researcher is required, the policy makes note of the unpredictable nature of genetic discoveries and the limitations of researchers to address some post-study issues. The NSERC policy statement provides that, "[t]he prudent researcher cannot assume that he or she can anticipate all harms inherent in a particular project."[242] Finally, the policy statement highlights the issue of ethnicity as it relates to genetic inheritance and counselling by recognizing that people define themselves differently in terms of biological, social, and cultural relationships. As a result, when researchers propose studies of "ethnically related genetic changes," the policy makes the vague warning that the researcher should "understand this issue and be able to provide the necessary counselling."[243]

As these policies and guidelines demonstrate, the rationales for genetic counselling are shaped by values – whether in the form of individual beneficence, individual non-maleficence, consideration of social benefit, social justice, or societal eugenics.[244] At present, professional guidelines ensure that, "[a]lthough there is no single ethical issue that unifies the field of genetics, informed consent, confidentiality and the potential for social harm and psychological distress are issues that physicians involved with testing should understand."[245] An important objective within much genetic counselling at present is the distancing of the beliefs and morals of the health care professional. This is attempted to ensure patient autonomy within assisted reproductive decision-making processes. The extent to which this communication of information is carried out in an objective, "non-directive" fashion is believed to be integral to the presence of "real" choices for the patient(s). It has been said that "[o]ur society's belief in the notion of patient autonomy, or the right of self-determination, has never been stronger."[246] However, the privileging of individual autonomy within genetic/reproductive decisions has the potential to reproduce tensions related to notions of "justice." Justice, in this sense, requires the examination of our approaches to genetic testing *from the perspective of people with disabilities.* From this perspective, we may rethink the degree to which we circumscribe individual choice (as we have begun to do by attempting to restrict sex selection, and prenatal testing for superficial traits) to attempt to preserve human diversity and the inherent value of people with disabilities. Caulfield cautions us about the practical difficulties associated with non-directive genetic counselling. He states:

> ...[w]hile a non-directive approach to counselling is undoubtedly the ideal, on a practical level it is arguably becoming difficult and unrealistic. Moreover, the probabilistic nature of

> genetic factors in common diseases, like cancer and heart disease, will further undermine the goal of nondirectiveness. Factors such as the socio-economic status of the patient, the perceived severity of the relevant condition, the reason for the genetic test and the willingness of the patient to act on the information, affect the "directiveness" of the counselling process. Notions of relational ethics, whereby counselling is viewed as a mutual exploration based on the goal of enabling individuals to enhance and enrich human relationships, cast further doubt on the paramountcy of individual autonomy as embraced by non-directive counselling.[247]

As a matter of principle, Caulfield argues that "nondirectiveness" should co-exist with an explicit recognition of the unique and individual context that exists in any counselling situation.[248]

## 6.3 Law Affecting Genetic Counselling

### 6.3.1 Abortion

> Probably the most harrowing, urgent, and complex counselling situation occurs when a serious disorder in a fetus is discovered by prenatal diagnosis. The couple must decide between having a child with a disabling condition, often of unpredictable severity, and terminating the pregnancy, a distressing and grievous experience. Parents must weigh many imponderables, and must sort out their feelings (that may differ), and choose between undesirable options – all in a limited time. They must appreciate the nature of the disorder, the range of severity, the sequelae of abortion, and the support services available. Some advocates for the disabled feel that fully informing the couple should include special efforts made on behalf of the fetus, such as emphasizing the availability of support services, or arranging visits with parents of children with the same disorder. This is sometimes done in special circumstances, but some counsellors feel that it can be taken too far, and could cause undue stress to couples who have made up their mind to abort. There is no consensus, and it would be useful to know more about what is being done in this context.[249]

This quotation attempts to display the complexity of genetic counselling in relation to pregnancy termination. It raises questions about what counselling should involve, about whose concerns should be paramount within the process, and about the way society views persons with disabilities. It is obvious that there are conflicts between those who view the potential of having a child with a disability as reason for termination of a pregnancy, those who advocate against termination on the basis of "disorder," or "disability," and those who may also have moral or religious convictions regarding abortion. It raises important questions about the extent to which prospective parents understand the realities of life for children with disabilities and their families, and about the extent to which genetic counsellors ought or ought not to provide access to such experiences. Notwithstanding these unresolved issues framed within the genetic counselling context, the law regarding abortion in Canada is clear.[250] The criminal law in Canada cannot be used to regulate a woman's access to abortion.

Prior to the 1988 Supreme Court decision in *Morgentaler*[251], section 251 of the *Criminal Code of Canada* prohibited abortions – unless performed in an "accredited" hospital with the approval of a therapeutic abortion committee. These requirements made abortions unavailable in many hospitals. Based on the principles of fundamental justice, the Court held that, "[s]ection 251 clearly interferes with a woman's physical and bodily integrity. Forcing a woman, by threat of criminal sanction, to carry a foetus to term unless she meets certain criteria unrelated to her own priorities and aspirations, is a profound interference with a woman's body and thus an infringement of security of the person."

Since 1988, however, there have been numerous attempts to restrict women's rights to abortion. These attempts have been based upon the notion that a foetus has, or ought to have, legal rights. In 1989, two cases were decided regarding attempted injunctions preventing women from legally terminating their pregnancies. In both *Murphy* v. *Dodd*,[252] and *Tremblay* v. *Daigle*,[253] the courts upheld the ability of women to seek abortions, denying claims by the (potential) fathers. In *Daigle*, Justice Dickson ruled that under the *Quebec Charter*, a foetus is not considered a "human being," and therefore it has no legal rights. In addition, the Court denied the argument that the father's interest in a foetus he helped create gives him the right to veto a woman's decision with respect to the foetus she is carrying.

In 1990, then Prime Minister Kim Campbell attempted to reinstate criminal law control over abortions. *Bill C-43* was introduced to restrict the availability of abortion. The bill survived the vote within the House of Commons on May 29, 1990, however, it was defeated in Senate (43 to 43) on January 31, 1991. In the 1996 case of *R.* v. *Lewis*[254], the Supreme Court of British Columbia upheld women's rights to a "free and safe" abortion as outlined in the *Access to Abortion Services Act.*[255] Justice Saunders ruled that "health care has fundamental value in our society. A woman's right to access to health care without unnecessary loss of privacy and dignity is no more than the right of every Canadian to access health." Most recently, in a 1997 case, *Winnipeg Child and Family Services* v. *G. (D.F.)*,[256] the Supreme Court ruled that a foetus *is not a legal person* and that the state is not entitled to take action against a pregnant woman on its behalf.[257]

*R.* v. *Drummond*[258] was a criminal case in which a pregnant woman inserted a pellet rifle into her vagina approximately 48 hours before the birth of her child, and fired a pellet into the foetus' brain. The pellet had to be surgically removed approximately 88 hours after the birth of Johnathan Drummond to preserve his life. Since the case involved the attempted "destruction" of a foetus, the court commented on matters related to abortion. The question before the Ontario Court (Provincial Division) was whether a motion to quash the charge of attempted murder against Brenda Drummond under section 223(2) of the *Criminal Code* which provides that an accused commits homicide when he/she causes injury to a child before or during birth, resulting in the death of the child after he/she becomes a human being should be successful. The decisions in *Daigle* and *R.* v. *Sullivan*[259] were followed. As such, the court confirmed that a foetus is not a "human being," but a part of a woman until its *live* birth. The court granted the motion on the basis that the child was born alive, notwithstanding the fact that he remained alive as a result of medical intervention. The court felt that if the "born alive" rule is troublesome for Canadians in light of medical advances which can be used to avert the death of a child injured *in utero* (resulting in a lack of criminal charge/sanctions for those who do so), change is a matter best left to the legislature.

Access to abortion is considered a health matter – between a woman and her doctor. In practice, however, it is highly unusual for abortions to be performed beyond 22 weeks gestation in Canada. As a result of the absence of criminal prohibition of abortion in Canada, there are no accompanying statutory provisions that permit abortion on the basis of "special circumstances" (such as in cases where a "serious defect" is detected in a foetus) after 22 weeks gestation.[260] However, international abortion legislation, such as that of the United Kingdom, permits abortion after the specified time-period of gestation (in this instance, 24 weeks), if "there is a substantial risk that if the child were born it would suffer from such physical or mental abnormalities as to be seriously handicapped."[261] Disability advocates are opposed to this position, as they assert that it does not result in equal treatment of disabled people. They state:

> ...the Section is offensive to many people; it reinforces negative stereotypes of disability; and there is substantial support for the view that to permit terminations at any point

> during a pregnancy on the ground of risk of disability, while time limits apply to other grounds set out in the Abortion Act, is incompatible with valuing disability and non-disability equally.[262]

## 6.4 Legal Principles

### 6.4.1 Standard of Care

The creation of a patient-physician relationship leads to the existence of a duty of care owed by the physician to the patient. This relationship obliges the physician to exercise a reasonable degree of care toward the patient.[263] Elements of a duty of care commonly include, "the duty to attend, the duty to diagnose, the duty to treat, the duty to refer, the duty to monitor, and the duty to provide appropriate aftercare."[264] When medical practitioners' actions fall below the accepted standard of care, they can be found liable for medical malpractice. "In general, to establish a case of medical malpractice, a patient must prove there was a duty of care, a breach of that duty, an injury, and a causal connection between the breach and the ultimate injury."[265] The duty of care owed to a patient by a physician requires specific articulation in the context of genetics.

> Genetic testing has the potential to create unique liability issues for medical genetics professionals. For example, emerging genetic technologies have the potential to affect the legal standard of care and the scope of the health care professional's duty to inform and duty to follow-up.[266]

It is clear that physicians do have a legal obligation to inform patients of the availability of prenatal testing.[267] A physician, however, is not legally obligated to take on a patient or provide health care services that conflict with personal moral convictions.[268] Thus, proper genetic counselling often requires referral to a professional team, to a genetics centre, or to a patient support organization.[269]

Genetic information is complex, and often contains a high degree of uncertainty. Most often, it also points to the (potential) existence of a trait or condition for which treatment is not yet available – this creates real questions as to how far a physician's duty to inform should extend.[270] At present, there is no clearly articulated *legal* standard of care for clinical genetic practice – and, the testing and counselling programs that are offered vary among provinces.[271] However, genetics are often considered a specialty within the field of medicine. "There is an obligation to refer a patient to a specialist when the health care professional lacks sufficient expertise."[272] Physicians (such as obstetricians or gynaecologists) who choose *not* to refer their patients may be understood to be representing themselves as competent geneticists, and as such, may be held to the standard of care of a geneticist.[273] It is argued that there is a specialized standard of care for medical genetics.[274] Sharpe asserts that on the basis of professional accreditation standards that require the ability to "communicate at a level appropriate to the [patient]" and "to provide psychological support," a geneticist's duty of care, (although analogous to that applicable to a physician) is clearly distinct.

A distinct (conceptual) model of a genetic counselling duty of care has been developed and applied to genetics by health care practitioners in Canada. Sharpe describes the model as emphasizing patient-centred "informed autonomy" which takes into account the communicative and psychological difficulties associated with the results of genetic testing.[275] The elements of this duty of care, as articulated by Sharpe include: the duty to take a detailed medical and family history; the duty to provide psychological support;

the duty to facilitate effective communication; and, the duty to monitor, provide appropriate aftercare, and not abandon.[276]

Genetic counselling must be provided (preferably by individuals well trained in dealing with genetic issues) prior to and following genetic testing.[277] It follows that they also have a legal obligation to take steps to ensure that the patient understands the information provided to her/him/them.[278] Sharpe argues that the psychological effects – upon partners, spouses or family members – must be taken into account by the physician to protect patient autonomy, since psychological difficulties can impair "communication, comprehension and adjustment."[279] Sharpe demonstrates the importance of communication for psychological well-being in the genetic counselling context:

> ...a patient's emotional and psychological responses to genetic information can significantly impair a patient's ability to comprehend and retain genetic information. Although these types of responses may prove to be nothing more than part of the normal coping process, adequate time must be provided for the patient to proceed through such reactions. Premature discussion of genetic information may lead to misunderstanding, dissatisfaction and rejection of counselling and aggravate a patient's feelings of anxiety and despair.[280]

Of the utmost importance within the "genetic model" is what is referred to as "informed autonomy" – in which empathetic education, communication and patient comprehension are facilitated.[281] Thus, any number of "psychological" difficulties that could impair the reproductive decision-making process must be taken into account by the physician to avoid "harm."

"From the perspective of good patient care, the standard of disclosure for genetic testing may be regarded as embodying the pre-test education, communication and psychological counselling processes."[282] Although it may be difficult to clearly articulate the responsibilities of a genetic practitioner prior to genetic testing and counselling, Sharpe suggests that this is part of the duty of care. He notes the opinion of those who suggest that by attempting to clearly articulate and live up to these responsibilities, genetic practitioners may be able to avoid regulation from outside the profession to which they are "unlikely" to adhere.[283] The discussion of all types of information – medical and non-medical – will be different for each patient within the genetic counselling process. To be assured that a physician is fulfilling his/her duty of care to the patient, this transfer of information must be non-directive to ensure that the ultimate decision reflects the wishes of the individual patient(s)/person(s) making reproductive decisions. This model exists in opposition to the paternalistic notion that the "physician knows best." However, critical communicative, educational, and counselling processes ensure that particular care is given to any discussion of risk information so that *each patient* is given adequate time to become aware of, to cope with, and to adjust to genetic information.[284]

> If physicians are to provide benefit and prevent harm to the patient before, during, and after genetic testing, they will have to make a fundamental change in the way they perceive the nature of their relationship with the patient, and how they can best respond to the patient's pleas for assistance and relief.... Given the lack of effective treatment and cure for many genetic diseases, physicians will have to focus on a patient's communicative, emotional and psychological needs in order to assist the patient to understand, adjust to, and cope with the implications posed by genetic testing and test information. In short, physicians will have to apply genetic counselling's distinct therapeutic model of care, and develop a "human vision" as opposed to a purely "medical vision."[285]

The "therapeutic" model of care attempts to avoid the demarcation of the beliefs of either the patient or the physician as "irrational." In this way, genetic counselling is intended to bring about decisions best suited for the individual(s) in question in order to avert "harm." In the context of prenatal testing, it is often difficult to allay anguish and dismay, as many believe that the genetic information obtained through testing presents, "no desirable options."[286] Proponents of the therapeutic model resolve this conflict by relying on the primacy of autonomy in decision-making. In the process of genetic counselling then, both parties must "willingly remain exposed to such risks, to trust in the uncertain, for these are the risks inherent in any autonomous decision."[287]

Another important aspect of genetic practice is confidentiality of information. Genetic information however, may be seen to differ from previous forms of medical knowledge on the basis of its ability to enable us to predict future health. Thus, it is not only relevant to a particular individual, but possibly to spouses, partners, and family members.[288] This raises important questions about the duty of confidentiality held by genetic professionals. Knoppers illustrates these questions. She states:

> ...genetic information is not only personal, but inherently familial. For example, a genetic test of one individual may have relevance to all blood relatives, or may reveal information about another family member that he or she does not know themself. Consequently, even the right to the confidential treatment of medical information – a basic right in the health care context – must be re-examined in cases involving genetic information.[289]

There are no statutory provisions governing the disclosure of genetic data. Currently, there is no legal duty to inform at-risk family members.[290] Genetic information places physicians in situations where, "the physician's duty of confidentiality will frequently clash with the physician's duty to prevent harm."[291] The question of whether disclosure of genetic information to family members can in some cases be either permitted without patient consent, or even required by law, begs investigation – by either the courts, or the legislature.

As part of their duties of care, generally, medical professionals owe their patients a duty to keep medical information confidential.[292] This duty is neatly summarized in the following way:

> [C]onfidentiality protects a patient's personal autonomy and privacy, and is meant to encourage the free flow of information between patient and medical professional, so that the patient may be provided with optimal care. Medical professionals may disclose information only if the patient has expressly or impliedly consented to the disclosure, or the law requires or permits disclosure notwithstanding the patient's refusal to consent. The law may require such disclosure in at least three circumstances: giving testimony in a court of law; under a statutory obligation, such as the reporting provisions in public health legislation; or in other circumstances where disclosure is required to protect a paramount public interest.[293]

The specific legal articulation of this duty is complex. It has arisen through tort, contract, and equity case law, statutes, and the ethical codes that govern health professionals.[294] "Medical professionals are generally obligated by law and professional ethics to hold in confidence information obtained about their patients, *including any genetic data.*"[295] This obligation arises from the physician's fiduciary obligations to the patient[296], and from various ethical codes.[297] Medical professionals who breach this obligation may be liable for damages flowing from the breach, and may also be subject to professional disciplinary proceedings. Tigerstrom et al. also suggest that it is possible that health care institutions, such as hospitals, cancer boards or regional health authorities, could be held to be in a fiduciary relationship with patients.[298]

Exceptions to a physician's duty of confidentiality arise both as a result of statute and the common law. They arise in situations of immediate and genuine risk of harm. Flanagan asserts that common law exceptions to the duty of confidentiality might arise in the context of genetic information. He asserts:

> ...[e]xamples of required or permissible statutory disclosure generally involve circumstances where there is a genuine and immediate risk of harm to members of the public, and the legislature has determined that the risk of harm outweighs the patient's right of medical confidentiality. This rationale has not yet prompted any jurisdiction to impose statutory privileges or duties with respect to the disclosure of genetic data to family members or reproductive partners. Therefore, the various common law exceptions to the general obligation of medical confidentiality may be of greater importance in the area of genetic data.[299]

*McInerney* v. *MacDonald*[300] is the leading Canadian case regarding the confidentiality of medical information. In this case, the court recognized that there is a fiduciary relationship between physicians and patients that obliges the physician to hold information about his or her patients in confidence. This obligation was found to be absolute unless, "the safety of individuals or of the public, physical or moral, would be sufficiently cogent to supersede or qualify the obligations *prima facie* imposed by the confidential relation."[301] Although there are no Canadian cases where liability has been imposed on a physician for failing to warn an identifiable third party of the risk posed by a patient, there is a body of law that indicates that a physician *may* be liable for foreseeable injury caused to identifiable third parties by a patient.[302] In future cases involving genetic data, the courts will have to balance the patient's right to medical confidentiality, and the biological relatives' interest in obtaining significant and useful genetic data.

While the confidentiality of personal health information is both a legal and ethical right, in the context of paediatric genetics it may be difficult to apply and uphold. "The inherently familial nature of genetic information coupled with an increasingly family-centred ethic of decision-making for children are factors that give rise to special challenges in the respect for and maintenance of confidentiality of the genetic information of a child."[303] The free flow of information between the health care provider and patient is essential to effective medical treatment. Indeed, in a recent decision concerning equality rights, the Supreme Court of Canada referred to "effective communication" as an integral and indispensable component of medical care.[304]

### 6.4.2 Consent

A physician's duty to disclose material risks arises from the duty of care in treating the patient. "The function of professionals possessing medical knowledge is to provide information that individuals consider material to their exercise of choice."[305] Thus, a patient's consent must be obtained prior to the performance of medical treatment. Historically, consent created a full defence to charges of assault or battery.[306] More recently, however, the Supreme Court of Canada has located the duty to inform for consent within the law of negligence.[307] Every mentally competent person has the right to refuse medical treatment – even if that treatment would be in that person's best interest. The right to refuse treatment is protected by the *Charter*.[308] To be valid, a person who has the capacity and who has been informed of the basic nature and consequences of the treatment must give consent voluntarily.[309]

"Informed consent" refers to the duty of a physician to disclose, "material information concerning proposed treatment prior to obtaining the patient's consent."[310] The goal of informed consent is to enable the patient to have the information necessary to make an informed choice. To give a patient relevant information, a physician must first ask relevant questions.[311] Failure of the health care practitioner to provide relevant information to the patient may give rise to an action in negligence against the practitioner. The leading Canadian case on medical negligence is the 1980 Supreme Court of Canada decision in *Reibl* v. *Hughes*.[312] *Reibl* changed the standard of disclosure from that of "what a reasonable patient would want to know," to that which "a reasonable physician would disclose." A reasonable patient, according to the unanimous court, would want to be informed of *all material risks* of a proposed treatment, as well as any *special or unusual risks.* "Material information" has since been defined by the courts in very broad terms and has been held to include both medical and non-medical consideration (e.g., the risks of side effects and the potential social ramifications of a treatment choice).[313] *Reibl* also restricted the tort of battery to cases where surgery or treatment is performed without any consent, or beyond the consent given.

As a result of *Reibl,* a health care provider must provide the patient with any information that a reasonable person in the patient's position would want to know. Knoppers asserts that in the case of genetic testing, this duty includes, "informing the patient of the nature and consequences of testing, material risks, and alternatives."[314] However, she also notes the difficulty associated with the communication of complex genetic information. She cautions that

> ...[m]uch of the information which must be relayed to the patient is probabilistic and couched in the ambiguous language of "susceptibility," "risk factors," and 'late-onset." In the absence of adequate medical and public education, this information could have little practical meaning to the patient, thereby undermining a physician's attempts to meet the expected duty to inform.[315]

With respect to genetic counselling, the requirements for informed consent are, or ought to be, distinct. Caulfield explains that

> ...[a] dynamic, responsive and ongoing consent process is required. Perhaps more than in any other area of medicine, the ethically and socially complex and rapidly advancing area of genetics requires that informed consent be a process where health care provider and patient candidly explore treatment options, patient rights and obligations (perhaps to third parties such as family members) and the future implications of testing and banking. Furthermore, because of the potential for subtle coercion, and for involvement over a number of years, the voluntary nature of testing should be continually underscored.[316]

Informed consent is founded largely on the ethical principle of autonomy. The Ontario Court of Appeal in *Mallete* v. *Shulman*[317] articulated that "[i]ndividual free choice and self-determination are themselves fundamental constituents of life." In *Ciarlarliello* v. *Schacter,*[318] a 1993 case of the Supreme Court of Canada, the court established that there is an onus on physicians to show that a patient understood the explanation and instructions that were given in order to protect the autonomy of the patient.

The protection of autonomy in health-related decision-making becomes difficult in the context of newborn screening for genetic diseases. Autonomy, in this instance, refers to a parent's ability to make health care decisions for her/his children. Since minor children are unable to give consent, questions arise as to when parental consent is required for participation in such programs. "In general, mandatory newborn screening programmes are for conditions of high incidence that have an accurate test and treatment capable of preventing the expression of the disease."[319] Thus, testing performed within a mandatory screening program does not require parental consent. This is justified by the notion that "the child's best interests outweigh parental values where the child's life or health is at risk."[320] Parental autonomy in decision-making is overridden to protect "the right to health of a child" in these instances. Parental consent is required however, in cases where tests are not as accurate, and treatment is not available.[321] However, as genetic medicine advances, and certain genetic tests become viewed as routine, the consent process can be undermined. Caulfield cautions that "[e]ven in the rare circumstance when testing may be viewed as justifiably mandatory, such as with neonatal screening programs for treatable diseases, the nature and implications (including psychological and socioeconomic) of the genetic test and the alternatives should still be fully explained to the participants (e.g., the parent(s))."[322] This also holds true in the prenatal testing and counselling context.

When prenatal testing occurs, the genetic counselling process must assist prospective parents to make extremely difficult decisions, based upon the ways in which the parent(s) *perceive* the "risks" at hand. Again, individuals are often unable or unwilling to address or communicate their understandings of such "risks." "Risk," in the prenatal context is more opaque and arguably, more subjective.

At common law, the standard of disclosure articulated in *Reibl* is a modified objective standard. However, within current genetic counselling practices, a "highly subjective" standard of disclosure is required by the objective of "non-directiveness." "The health care provider should communicate at a level *appropriate to the patient.*[323]

According to Sharpe, the "genetics model of informed consent" is distinct from the model currently applied by Canadian courts. The standard for disclosure in genetic testing and counselling is very subjective due to the nature of "risk" involved. It is very difficult for physicians to intuit which information a particular patient desires, or does not understand. When interpreted legally, in light of the *Reibl* disclosure standard, this highly subjective communicative process could be controversial, or difficult to reconcile. Given the nature of the skills necessary to communicate effectively, and the nature of the information to be discussed, Sharpe argues that in the context of genetic testing, "[m]any physicians do not understand that the recommended standards of communication, counselling and informed consent represent a significant departure from the standard of care generally applicable to physicians." Particularly in instances where treatment does not yet exist for particular conditions identified by genetic testing, "material information" takes on special significance. Sharpe asserts that

> ...instead of providing information concerning a proposed treatment and the material risks associated with it, material risks must be assessed with respect to the impact of receiving test results. A patient's informational needs and decision will be dependent upon, and shaped by, the patient's own objectives, concerns, values and beliefs. This is information that a patient may be unwilling or unable to discuss with the physician. Accordingly, a physician as well as a court of law, may find it difficult to determine what a "reasonable patient" would want to know. This raises the question whether the Reibl v. Hughes test for assessing materiality is applicable, or appropriate, within the context of predictive, carrier and population genetic testing, and good patient care.[324]

### 6.4.3 Causation

In all negligence cases, "the plaintiff... bears the onus of proving (on a balance of probabilities) that the doctor's negligence was the cause of the plaintiff's injury."[325] "Cause-in-law is determined by asking if the undisclosed information had been provided, would the patient have consented to the treatment? If the answer to this question is no, and cause-in-fact has been proven, the plaintiff will recover damages for the injuries."[326] Robertson argues that the 1990 Supreme Court decision in *Snell* v. *Farrell*,[327] in which Sopinka J. referred to the need for a "robust and pragmatic approach, has had the effect of "making it easier for plaintiffs to prove causation in malpractice suits."[328] This, he says, is based on the decision to allow evidence to permit an *inference* to be drawn that the doctor's negligence caused the plaintiff's injury.[329]

There is much dissatisfaction with the standard of causation applied in medical malpractice cases in Canada. It has been suggested that the legislature take action, and, "intervene to create a recovery mechanism for patients whose interests have been violated by their doctor's negligent failure to disclose a risk." However, at present the standard as to what risks must be disclosed should remain that outlined by Laskin C.J. in *Reibl – all material and unusual or special risks."* [330] The Supreme Court in *Reibl* applied a modified-objective standard of causation to avoid actions by plaintiffs acting upon "hindsight and bitterness."[331] Thus, the question was whether, "a reasonable person in the plaintiff's position would not have consented had the information been disclosed."[332] Sharpe explains the perceived effect of this standard as

> ...[t]he modified objective test for causation seemingly recognized the significant role a patient's particular concerns and objectives can play in medical decision-making. As such, the test was initially perceived as a rejection of the professional medical standard. Some have argued the modified objective test for causation effectively could subordinate a plaintiff-patient's concerns, values and objectives to their medical counterparts in the form of expert medical evidence concerning what decision a reasonable person in the patient's position would make.[333]

Although Sharpe argues that a subjective standard of disclosure should be applied to informed consent in the context of predictive genetic testing (and arguably, genetic counselling), he concludes that it is less clear whether a subjective test should be applied for causation. In the context of prenatal testing and genetic counselling, Sharpe concludes that the following standard should be applied:

> ...[t]o establish a causal link between the physician's failure to disclose information and the plaintiff-patient's injury, the court should inquire whether the health care provider undertook all reasonable procedures to facilitate effective, empathetic communication and comprehension "in a way that is meaningful to the individual or family", and "at a level appropriate to the consultant [the patient].[334]

Such "procedures" can include:

> ...providing appropriate time to facilitate an effective communicative relationship, the timing, the manner, the length and the place of all discussions, the impact of emotional and psychological responses and a consideration as to who provided the information, taking into account any cultural, socioeconomic and linguistic differences which may impair a patient's understanding.[335]

Sharpe acknowledges the practical difficulties of such communication. However, he concurs with those who argue that "...a practical response may be the application of health care personnel who have specialized communicative and emotional/psychological counselling skills, such as genetic nurses."[336]

## 6.5 Tortious Liability

The concept of a duty of care owed to the foetus is "well-entrenched" in Canadian law. This forms the basis of liability for negligence causing prenatal injuries. In an action for medical negligence, a plaintiff/patient must prove: the existence of a duty of care; breach of that duty; and, a causal link between the defendant's negligence and the plaintiff's injury.[337]

Although we have yet to see many cases litigated within the context of genetics, some presume that liability will likely be extended "for negligent acts committed when the child was not yet conceived, for example in the context of pre-conception genetic counselling."[338] Awarding damages for children born with "disabilities," or genetic "disorders" implies that "disability" can be construed as a "harm" that is either the result of a negligent commission – such as a "botched" abortion procedure, or an omission, such as the failure to inform a patient of a serious "risk." Often the "harm" resulting from the (mis)communication of genetic information by genetic counsellors or practitioners is nearly impossible to avoid since neither the termination of pregnancy, nor the birth of a disabled child is perceived as a desirable option. In this instance, the resulting psychological impacts often cannot be reversed. Damage awards, as is their nature within tort law, are intended to "fix," or place an individual in the situation he or she would have been in had the negligent events not occurred. In the context of disability, "repair" is not only often impossible, but in addition, monetary compensation for "disability" reinforces the view of people with disabilities as "unhealthy," "abnormal" or in need of repair or improvement.

### 6.5.1 Prenatal Liability

In most negligence actions, the existence of injury is readily apparent. However, in cases alleging prenatal injuries, courts often have a more difficult time determining whether an injury was sustained.[339] "Prenatal injury" actions involve claims by (or on behalf of) a child born in a "damaged" condition alleging that its injury was caused by the negligence of another before its birth. The claims are for the losses associated with being born "injured" where the injury is the result of someone's negligent conduct. In the 1933 Supreme Court case of *Montreal Tramways* v. *Leveille*[340], a pregnant woman was injured by a tramcar owned by the defendant. Her child, who was born with clubbed feet two months later, brought an action for the prenatally sustained injuries that caused the damages. The Court held that the child had the right to sue. More recently, in *Duval* v. *Sequin,*[341] a negligent driver caused a traffic accident that resulted in serious injuries to a pregnant woman, and to her foetus. The severe disabilities of the child bringing the claim were solely attributable to the accident. Following *Donoghue* v. *Stevenson,*[342] the Ontario High Court found that an unborn child is within the foreseeable risk incurred by a negligent motorist, and awarded damages. It held, "[w]hen the unborn child becomes a living person and suffers damages as a result of prenatal injuries caused by the fault of the negligent motorist the cause of action is completed. A tortfeasor is as liable to a child who has suffered prenatal injury as to the victim with a thin skull or other physical defect."[343] In these cases, the problem that the injured party is a foetus is solved by the crystallization of the cause of action when the child is born alive.[344]

Injuries to a foetus may also occur during gestation as a result of the conduct of its mother – such as, the use of drugs, tobacco or alcohol, or other negligent behaviour.[345] Causes of action resulting from such behaviour rest upon the finding of a duty of care owed by a mother to her foetus breached by the mother's conduct. Canadian case law relevant to this issue involves both public, and private responses to this notion of a maternal duty of care. The state has often acted to impose conditions of treatment upon pregnant women assumed to be harming their unborn children through their abuse of alcohol, drugs, or other noxious substances. Similarly, actions have arisen by (or on behalf of) children against their mothers alleging negligent behaviour.

The case of *Winnipeg Child and Family Services* v. *G.(D.F.)*,[346] proceeded despite a recommendation of the Royal Commission on New Reproductive Technologies (RCNRT) against judicial intervention during pregnancy (gestation and birth).[347] Although decided in the context of the powers of the state to interfere with a woman's pregnancy, the case illustrates the hesitancy of the courts to place the interests of the woman and her foetus in conflict – the result being the loss of autonomy of the woman.[348] In *G.(D.F.)*, the Manitoba Court of Appeal overturned the decision of the trial court to exercise its *parens patriae* jurisdiction to grant an injunction requiring a pregnant woman with a history of solvent abuse to commit her to custody, and enter a treatment program. The argument that the mother's allegedly tortious behaviour ought to be restrained was rejected. This speaks to further claims against a mother by her child for damages resulting from prenatal injuries. The court held that the conflict between the rights of the mother and those of the child should be resolved *in favour of the mother*.[349] The Supreme Court of Canada upheld this decision.[350] The decision of the majority was based largely upon the proposition that any rights or interests a foetus may possess remain incomplete until birth. As such, an action could not be brought on behalf of the foetus to restrain the mother from potentially harmful actions. The Supreme Court confirmed that the *parens patriae* jurisdiction of the court does not extend to unborn children. Although the issue of whether a mother owes a duty of care to her unborn foetus was raised, it was not addressed by the court other than to state that even if there is a duty of care that could give rise to a claim on the part of the child once born alive, there is certainly no such right in the case of a not-yet-born foetus.

*G.(D.F.)*, when viewed in light of emerging genetic practices that, arguably, have the eradication of disability as their objective, illustrates the tenuous relationship between one prevalent view of abortion and disability. The dissent in *G.(D.F.)* relied on the belief that state intervention in pregnancy is justified. Rather strange logic was followed within the dissenting opinion that stated that if women wanted to avoid state intervention in their pregnancies, they could do so by "choosing" to have a state-sanctioned abortion. More telling is the rationale behind such potentially court-sanctioned interventions. Major J. stated that intervention "serves the interests of mother and foetus and prevents unnecessary spending by the Canadian government to care for children born with disabilities."[351] He also suggested that a tort action may be possible by the child against her mother for "lifestyle choices," where the necessary elements of a negligence claim can be proven.[352]

*Dobson* v. *Dobson*[353] is the first Canadian case in which the court was confronted directly with the question of whether a mother should be liable to her child for injuries that they received in the womb as a result of her negligent actions. While driving, Cynthia Dobson was involved in a collision. At the time, she was twenty-seven weeks pregnant. That day, she gave birth to her son Ryan by Caesarean section. Ryan suffered permanent physical and mental impairment, including cerebral palsy. It was alleged that his mother's negligent driving directly caused Ryan's conditions. At trial, Ryan was found to have a cause of action against his mother thereby allowing the potential creation of a new duty of care between a mother and her unborn child.[354] The trial court relied on the fact that the common law has allowed a child, injured while a foetus *in utero*, to bring an action against a third party for his or her injuries to sustain an action. The New Brunswick Court of Appeal dismissed the appeal, and narrowed the inquiry to the specific circumstances surrounding pregnant women in the context of driving – as distinct from situations involving the "lifestyle choices" of pregnant women.[355] The Court of Appeal held that a child injured due to his mother's negligent driving should be able to sue for compensation. The Supreme Court of Canada allowed Cynthia Dobson's appeal, and held that *a mother is not liable to her children for prenatal injuries caused by her alleged negligence.*

The Supreme Court disagreed with the finding of the trial court that the success of negligence claims against a third party supports the creation of a similar duty between mother and unborn child. The majority did inquire as to whether a duty was owed by Cynthia to Ryan, as this type of examination was found to be against public policy. The duty of care was not extended to the mother for three reasons: because of the ensuing conflict it could create between a woman and her foetus; because of the difficulty of establishing the nature of "tortious" behaviour throughout the daily life of a pregnant woman; and, because certain behaviours (for example, alcoholism or drug addiction) are beyond a person's control and thus would not be deterred by the imposition of a duty of care. According to the majority, this type of duty could be created only by the legislature. The dissent preferred to keep the issue of negligence narrowly confined to the context of driving, and on this basis, asserted that this type of action should be permitted – even in light of the fears of the majority that it would lead to a slippery slope of maternal prenatal liability.

Currently, the common law position regarding the relationship between a mother and her unborn child is clear. The courts have *not* imposed a duty of care on pregnant women in order to preserve the autonomy rights of women, and to avoid making proclamations about the personhood status of the foetus contrary to those established within the laws and jurisprudence surrounding abortion.

### 6.5.2 Wrongful Birth/Wrongful Life

Medical malpractice liability is based on the concept of fault, or negligence. To obtain compensation for injuries sustained in the course of medical treatment, an individual must prove that the injuries were caused by the fault of the physician/health care professional.[356] As genetic counselling and prenatal diagnosis become more available, the issues of whether patients have been injured through the receipt of negligently delivered genetic services, or through the negligent failure to provide genetic services have arisen. New technology and knowledge create a space within law for the articulation of (potentially) new standards of care. Whether knowledge and "risk" were communicated appropriately, and whether "harm" was inflicted become increasingly vital issues. The introduction of new screening and testing technologies may raise the expectations of the parent or parents to have a child free of a genetic "defect." As Caulfield and Robertson note, "[t]he mere existence of wrongful birth and wrongful life claims may cause physicians to encourage the use of genetic services and to subtly recommend the termination of pregnancies which may result in the birth of a child with a disability."[357]

Wrongful birth and wrongful life actions share the claim that the defendant's negligence caused the birth of a child, which by itself, is an injury for which damages should be awarded.[358] To succeed, the plaintiff must prove that the health care professional acted negligently and that the negligent behaviour caused the harm.[359] These suits have greatly complicated the ways in which the courts address the issue of liability for malpractice by physicians. On one hand, their aim is to ensure that genetic practitioners are accountable if negligent, in order to provide compensation to parents for the costs of raising and caring for children with disabilities or to children for having to "endure" such disabilities. In this way, these cases resemble most other "ordinary" torts, whereby a finding of negligence and an award of compensation attempt to "return the injured person to the status quo, to deter negligent behavior, and to utilize insurance to spread the costs of accidents throughout society."[360] However, it is becoming exceedingly difficult to separate the fundamentally moral issues arising within these cases from those of the "regular tort." This is a result of the very questions about the *value* of particular lives (i.e. those of persons with disabilities) we are asking the courts to address. As Dworkin illustrates:

> Should a person ever be compensated for being born? Since the alternative to the birth of a child with a defect is often an abortion, how can the law compensate a person for being denied the information that would have led to aborting a pregnancy? And how can we make judgments about which lives deviate so disastrously from "normal" that a parent must be paid for having an "abnormal" child or the child must be paid for the wrong of having a life?[361]

The success of wrongful birth and wrongful life claims rest, in part, on how we as a society feel the individual, or the state should deal with the costs associated with caring for persons with disabilities. At the outset, this type of thinking is inherently undesirable as it immediately constructs disability as a problem that requires "special" or additional resources rather than as occurring within the natural spectrum of human difference and acceptance. Nonetheless, within the legal system, reliance upon the tenets of corrective justice attempts to resolve this issue by placing responsibility on the shoulders of the negligent health care professional.[362] However, due to the proven inefficiencies, expenses, and holdings for compensation for those who are legally entitled, it is also argued that alternative avenues of compensation – such as the creation of improved social welfare programs for families with children who have disabilities – must be pursued.[363]

Wrongful birth actions rest on the notion that a health professional deprived the parent(s) of a child of accurate information that would have "led them to refrain from having the baby in the first place."[364] Wrongful life actions, on the other hand, rest on the claim that "the counsellor (or health care professional) had inflicted the injury of being born on the baby, who, presumably, would have been better off never to have lived."[365] Each claim rests upon an assertion of medical malpractice based on a failure of communication (or action) regarding the risks of having a child with disabilities. At present in Canada, wrongful birth suits have been successful but wrongful life suits have not.

In wrongful birth cases, the plaintiff is someone other than the child – usually the child's parents. Two types of damages are generally claimed: financial and emotional. These actions are more likely (than wrongful life) to result in the awarding of damages since the courts are more willing to recognize the duty between the physician and the parent(s) of a child, than one between a (then unborn) child and physician.[366] Wrongful birth cases arise in varied contexts including: "failed medical sterilization or abortion, negligence in prescribing contraceptives, and negligence in genetic or other prenatal counselling or diagnosis."[367] Thus, the alleged negligence in such cases can occur before, or after conception.[368] These cases are perceived by some as being controversial because they (potentially) involve the awarding of damages for the birth of a "healthy" (i.e. non-disabled) child. The correlation assumed within such actions between "health" and "disability," however, is controversial in itself, as it implies that people with disabilities *are not* healthy.

In the late 1970s, Canadian courts resisted wrongful birth claims. This is illustrated in the cases of *Doiron* v. *Orr*,[369] and *Culp* v. *Ringrose*.[370] *Doiron* involved the sterilization of a young mother, in which a relatively uncommon, unsuccessful, yet recognized procedure was to be utilized. Finding that Mrs. Doiron had been sufficiently advised of the risks of pregnancy associated with the procedure, the Court dismissed her claim for what were termed, "extensive" damages.[371] *Culp* was a similar medical malpractice action in which Dr. Ringrose used an experimental agent to perform a sterilization procedure. Although the agent caused significant scarring to Mrs. Culp's fallopian tubes, accompanied by considerable pain and grief, the Alberta Supreme Court held that it was against public policy to award damages for the birth of a healthy child.

Less than ten years later, *Cataford* v. *Moreau*[372] began to change the landscape of wrongful birth claims in Canada, as the Court reconsidered whether it was still contrary to public order to grant damages following the birth of a healthy child. The language of the decision, however, signals the persistence of deeply entrenched, secular beliefs about abortion, and the "nature" of the family. The Court stated that "... the event [birth] constitutes a blessing intended by the natural order of things."[373] Nonetheless, both the wrongful birth and the wrongful life claims were addressed. Although the wrongful life claim was dismissed, the wrongful birth claim was successful. Damages in the amount of $2,000 were awarded for loss of enjoyment of life, the inconvenience and anxiety of the pregnancy, and the inherent suffering and inconvenience related to the operation required to properly execute Mrs. Cataford's sterilization. This case was followed by *Keats* v. *Pearce*,[374] in which the damages awarded to the successful litigant were limited by her failure to take "reasonable efforts" to mitigate her loss. Here, notwithstanding the finding of medical negligence after an improperly performed tubal ligation, "the court concluded that a reasonable step would have been to offer the child up for adoption, and thus it awarded only $4,200 in damages."[375]

Causation requirements in wrongful birth actions are based on a modified-objective standard of a "reasonable woman in the position of the patient." The courts must decide whether a reasonable patient would have terminated her pregnancy in light of genetic information, if given the option. The rationale and guidelines for the test as enunciated by the majority of the Supreme Court of Canada are as follows:

> The test... relies on a combination of objective and subjective factors in order to determine whether the failure to disclose actually caused the harm of which the plaintiff complains. It requires that the court consider what the reasonable patient in the plaintiff's circumstances would have done if faced with the same situation. The trier of fact must take into consideration any "particular concerns" of the patient and any "special considerations affecting the particular patient" in determining whether the patient would have refused treatment if given all the information about the possible risks. The "reasonable person" who sets the standard for the objective test must be taken to possess the patient's reasonable beliefs, fears, desires and expectations. While evidence of reasonable fears and concerns can thus be taken into account, purely subjective fears which are not related to the material risks should not be considered. The modified objective test strikes a reasonable balance, which cannot be obtained through either a purely objective or a purely subjective approach. A purely subjective approach fails to take into account the inherent unreliability of the self-serving assertion of a plaintiff, while the purely objective standard might result in undue emphasis being placed on the medical evidence, essentially resulting in a test which defers completely to medical wisdom.[376]

This test has proven to be "a formidable hurdle for patients to overcome when suing their doctors for negligently failing to disclose a material or unusual risk."[377] It is a highly problematic test in which many assumptions are made about "reasonable" beliefs regarding religion and abortion, and disability. However, the rationale for its use speaks to the "unreliable" nature of testimony within a purely subjective test – it is assumed by many that the plaintiff would always testify that the failure to warn was the determining factor in his or her decision to take the harmful course of action.

In both *Arndt* v. *Smith*,[378] and *Mickle* v. *Salvation Army Grace Hospital*,[379] the courts found that a reasonable woman in the position of the patient *would not* have terminated her pregnancy. *Arndt* dealt specifically with the issue of "undisclosed risk." Mrs. Arndt contracted chicken pox during her pregnancy, sought medical advise to attain knowledge of whether her foetus was at risk, and was told only of the least severe risks. Dr. Smith did not advise her of the more severe, but less probable risks of foetal injury as she did not want "to unduly worry an expectant mother about an improbable risk and one for which she would not advise therapeutic abortion."[380] The original claims in this case were made on behalf of Mrs. Arndt, her husband, and their severely disabled daughter – the latter two were dismissed. While attempting to apply the causation test in an objective fashion, the determination of causation relied upon factors such as: the stage of Mrs. Arndt's pregnancy with respect to the possibility of abortion, and of the risks of abortion at that time; whether a therapeutic abortion committee would have approved the abortion given the risks; whether the pregnancy had been planned, and the child wanted; and, Mrs. Arndt's feelings about the practice of medicine. On this basis, it was held "that Ms. Arndt would have chosen in foresight to carry Miranda to full term had she been fully apprised of the nature and probability of risks her foetus faced."[381] Notwithstanding her failure to disclose material risks, Dr. Smith was absolved of liability. Although the case was taken to the Supreme Court of Canada, the decision of the trial court was upheld.

Since *Arndt*, wrongful birth (and wrongful life) cases have arisen in the genetic context, and have dealt with negligence in failing to provide or to refer patients for genetic counselling, or in failing to inform parents of the availability of prenatal testing. According to Nelson and Robertson,

> ...[t]hese cases emphasize a legal principle which is now clearly established in Canadian law: a health care provider has a positive legal duty to inform the patient of *all material information concerning the existing (or proposed) pregnancy, including procedures which will test for possible fetal abnormality.* Failure to do so constitutes negligence.[382]

*H.(R.)* v. *Hunter*[383] involved a finding of negligence against two doctors for failing to recommend genetic counselling. As a result, two children were born with Duchenne muscular dystrophy. In addition to general damages, special damages were awarded for the cost of future medications and loss of future income for both the child's parents, as well as the costs of future care for the two severely disabled boys. The total amount of damages awarded was $3 million.

In *Krangle (Guardian ad litem of)* v. *Brisco,*[384] Drs. Brisco and Morrill failed to adhere to the generally accepted standard of care for a physician of a patient over the age of 35 who wishes to, or has become pregnant. This standard requires the physician responsible for prenatal care to advise her about and to offer to perform genetic testing.[385] If genetic testing had been performed, the results would have shown the presence of Down Syndrome. On the facts, the British Columbia Supreme Court found that given this information, Mrs. Krangle, as would most women in this position, would have opted to terminate the pregnancy. Following *Hunter*, damages were awarded for the loss of opportunity to have an abortion following failure to be provided with necessary medical information or advice. The Krangles were awarded the costs of caring for the child until the child reached 19 years of age, contingent costs for adult care (the remainder to be provided by the provincial welfare scheme), income loss, and non-pecuniary loss. *Krangle* helped to articulate the four general categories of wrongful birth cases: unwanted conception following failed sterilization; unwanted birth following failed abortion; loss of opportunity to have an abortion following failure to be provided with necessary medical information or advice; or physical damage to the foetus as a result of a medical procedure during gestation.[386] In reference to the quantum of damages to accrue in each category, the court offered limited guidance, stating that "[w]hat damages are recoverable and by whom (mother, father or child) depends upon the particular category into which the case falls and whether the child was born healthy or disabled [sic]."

After the trial, the support provisions of the British Columbia *Family Relations Act* were amended.[387] As a result of the amendments, parents of ill or disabled children over the age of 19 could be held responsible for the support of their child. The Krangles were concerned that if they could be found liable for their son's care after the age of 19, that the province would seek recovery from them for the costs of his adult care in a group home. On this basis, they argued that the award of damages for adult care should be increased. These issues were raised before the British Columbia Court of Appeal.[388] The majority of the court allowed the appeal, as it concluded that under the legislation, the province had an enforceable claim against the Krangles for the costs of Mervyn's adult care. The matter was referred back to the trial court for an assessment of the costs of adult care, at which time Dr. Morrill appealed to the Supreme Court to reinstate the trial judgment.[389] The Supreme Court restored the judgment of the trial court. The Court relied on the principle that damages must be assessed – once and for all – at the time of trial. It also held that although there are certain circumstances when parents can be held financially responsible for an adult "child," Mervyn Krangle, at age 19, once within a group home, would no longer be considered a child, "in the charge of his parents." Thus, the provincial social security program will still bear the majority of the costs of his care.

In *Patmore (Guardian ad litem of)* v. *Weatherston,*[390] an interesting decision by the Supreme Court of British Columbia, the court was faced with a counter argument by the physician accused of negligently failing to provide genetic testing. The physician argued successfully that the mother of the child bear responsibility for the birth of the child since her negligence in failing to attain and follow medical advice resulted in the birth of a child with Spina Bifida. This is referred to as a "wrongful birth" case because the court found that Ms. Blakeborough would have terminated her pregnancy had she known that her child would have Spina Bifida. However, her claim that the physician acted negligently was dismissed. After her first visit to Dr. Weatherstone, Ms. Blakeborough was informed specifically as to when the physician wanted to conduct a second ultrasound. She did not attend this appointment. The court found that had she attended this appointment, the second ultrasound would have confirmed the presence of the child's Spina Bifida and other disorders. However, the court did not place an obligation on the physician to "chase down" the expectant mother in this case, since at the time of her first visit, her pregnancy was determined to be without complication. Ms. Blakeborough argued that she was not properly advised regarding follow up visits; however, the court decided this not to be the case – particularly because she had previously had two other children. By the time of her second visit to the physician, the gestational age of the foetus she was carrying was over 19 weeks. At this time, Dr. Weatherstone did not order another ultrasound, although experts who testified at trial said that although it would have been difficult, a second ultrasound could have been arranged and carried out quickly. However, the court also considered evidence presented by Dr. Weatherstone's experts who opined, that at the relevant time since no foetal abnormalities were suspected, a second ultrasound was not necessary. Since this was not a case "fraught with risk," the court did not rule as to whether this situation was one in which the standard practice of the medical profession fell below the expected duty of care. The court stated that *had* it held that Dr. Weatherstone was liable for failing to perform a second ultrasound at 19 weeks gestation, liability would have been shared equally between himself and Ms. Blakeborough.

In wrongful birth cases involving children with disabilities, commentators note that the courts *have not* been reluctant to award damages – "even very substantial damages."[391] Damages, in this sense, are quantified on the basis of the costs of caring for a child with disabilities. However, the way in which these damages are calculated remains controversial. Nelson and Robertson speculate that Canadian courts will follow the example set by the House of Lords in cases where, notwithstanding the finding of negligence, a "healthy" (i.e. not disabled) child is born. In this instance, the courts have not awarded damages.[392] Where children are born with conditions resulting in disabilities however, damages attempt to compensate for the additional costs of raising the child. Questions regarding the duration of the compensation were addressed in *Krangle*, when the Supreme Court upheld the award of damages affixed by the trial court to compensate the Krangles for their son's expenses while he remained *a child in their care*. Questions regarding the degree of disability necessary to warrant damage awards remain, arguably, unanswered.[393]

In wrongful life cases, it is the child who brings suit and alleges that his or her own birth is an injury.[394] The "injury" claimed refers to the child being born with a disability, or as some have suggested, "unsatisfactory traits or prospects."[395] Thus, the child asserts a right "not to be born." As Robertson et al. explain:

> ...if an obstetrician negligently fails to detect the risk of fetal abnormality, and a child is subsequently born with disabilities, the child's wrongful life action would allege that but for the obstetrician's negligence the mother would have chose to have an abortion, sparing the child from a life of disability.[396]

The alleged negligent omission in wrongful life claims can occur prior to conception (i.e. failure to advise the parents of the risk of their future child inheriting a genetic defect; or the negligent use of a cloning technique to create a child); before implantation (i.e. negligent selection of a less than optimal embryo for implantation); or during gestation (i.e. negligent failure to advise the mother of the likely condition of her child).[397] In theory, wrongful life actions could be invoked to, "regulate the negligent use of any prenatal genetic diagnostic technique, some negligent uses of IVF, and even negligent attempts to create a child using a cloning technique."[398] In addition to the assertion that the child ought not to have been born is the claim that living with the disability has, and will continue, to cause pain and suffering and financial loss.[399] Thus, wrongful life claims can be advanced in concert with wrongful birth claims.

At issue in wrongful life cases is the principle of "harm." There are several competing conceptions of harm at play within wrongful life discussions. There exists the belief that harm should be measured or compared to the state of existence not characterized by that harm. In cases involving genetic testing, this is often understood to play out in either of two ways – a child is born with an impairment or disabling trait, or the pregnancy is terminated on the basis of information and counselling regarding the disorder or disability. In this sense, there can be no harm since the alternative is non-existence.[400] In the alternative, harm can also be defined as placing an individual in a position "in which that individual is disabled or suffering in some way or in which his interests or rights are frustrated."[401] This definition of harm does not involve comparison to an alternative, but accepts disability as an inherently undesirable and unworthy state of being. Still another conception of harm – one not related to a "wrong" perpetuated against a particular individual – situates harm within the context of the financial deprivation, and/or need of an individual born with disabling traits or conditions. "[I]n some circumstances, a wrong will be done to a child denied financial support where its [sic] condition is such that financial support is necessary for it [sic] to acquire or maintain these capacities for itself [sic]."[402] Although this definition of harm avoids philosophical as well as public policy pitfalls, it has the potential to place the "burden" or responsibility for caring for individuals with "special" needs upon individual members of the medical profession. Arguably this is a responsibility best left to state. Within our neo-liberal climate, characterized by both the depletion of welfare schemes and privatization of state services and responsibilities, it remains to be seen whether the courts will adopt this understanding of harm in the wrongful life context.[403]

The Canadian courts have not recognized the wrongful life claim. The onus in wrongful life cases is on the child to demonstrate that, had the proper procedure been provided, or had the abnormality been detected, the pregnancy would have been terminated. In the 1987 case of *Cataford* v. *Moreau*, the Quebec Supreme Court rejected the claim for wrongful life on the basis that it was contrary to public policy. In this case, a failed sterilization led to the birth of an unwanted, although otherwise "healthy" child. The Court maintained that

> ...[t]his claim is without any foundation. The birth of a healthy child does not constitute... damage, and still less damage compensable in money. It is clearly impossible to compare the situation of the infant after his birth with the situation in which he would have been in if he were not born. Merely to state the problem shows its illogicality; *by what perversion of the spirit may one arrive at qualifying as damage the inestimable gift of life?*[404]

The case of *Cherry* v. *Borsman*[405] differs factually from *Cataford* because a child was born with severe physical and mental disabilities including "brain damage, cerebral palsy, arthrogryposis and abnormal features." At trial, both the child and parent plaintiff were awarded damages as compensation for the trauma suffered during an unsuccessful abortion. The physician was found to have acted negligently both in his performance of the abortion procedure, and in his post-operative care. The British Columbia Supreme Court found that the physician owed a duty of care to both the mother and to the foetus. The duty to the foetus, however, was understood by the court to arise once the child was actually born. The court analogized the duty to that owed to children injured *in utero* by thalidomide. The right to claim for damages in such situations, said the court, accrues to the child upon live birth. The court determined this case on the basis of "ordinary negligence principles," rather than as a claim for wrongful life. The court found the physician liable for all costs associated with the birth of the child, and held that both the adult plaintiff and the infant plaintiff were entitled to recover for their pain and suffering and for their income loss. The court also held that the child was entitled to the costs of future care. The British Columbia Court of Appeal dismissed the appeal.[406] In *obiter*, the court also stated that the case could be tried successfully as a wrongful life action since the doctor admitted that he was negligent during his post-operative care and since the adult plaintiff would have had a second abortion, had she known of her continuing pregnancy earlier. Thus, since the child alleged that the negligent behaviour of the doctor led to her injury, the court was of the opinion that a wrongful life claim could, in theory, proceed.[407]

The way the trial court awarded damages in the *Cherry* case is somewhat confusing in light of the differences between wrongful life and wrongful birth claims. The award of future costs to the child resulting from care requirements is typical of a wrongful *life* action. However, the damage awards (here, to the mother *and* the child) for financial loss and emotional suffering is characteristic of a wrongful *birth* action. *Cherry* is said to be "very similar" to a wrongful life claim, although it has not been interpreted as such in later decisions. More recently, Canadian courts have continued the explicit rejection of wrongful life actions.[408] In *Arndt* v. *Smith*,[409] the claim for wrongful life was abandoned by the trial judge at the beginning of the trial as being unsustainable since wrongful life claims have no precedent in Canadian law and have been rejected in both England and the United States.[410] Similar public policy reasons were given for the dismissals of *Mickle* v. *Salvation Army Grace Hospital Windsor Ontario*[411], *Lacroix (Litigation Guardian of)* v. *Dominique*,[412] *Jones* v. *Rostvig*,[413] and *Patmore (Guardian ad litem of)* v. *Weatherston*.[414]

The most recent wrongful life case of note, however, is *Bartok* v. *Shokeir*.[415] Following the birth of their first child, the Bartoks sought the advise of their physician as to whether their son's condition, microcephaly (a shrunken, underdeveloped brain), was genetic in nature and whether their future children might also be at risk. The physician (a geneticist) advised them that it was not a genetic condition and that their future children had "zero risk" of being born with the condition. However, their second child was born with microcephaly as well. In addition to the claims of the Bartoks and their first child, the second child ("Kurt") also asserted a claim against the physician for economic loss resulting from the physician's negligent misstatement. The physician attempted to use Rule 173(a) of *The Queen's Bench Rules of Saskatchewan* to strike out the portion of the plaintiffs' statement of claim relating to Kurt. This provision, according to the court, can only be used "where it is plain and obvious that there is no cause of action." Based upon the facts of the case, and since Kurt's claim is for economic or pecuniary losses (rather than non-pecuniary losses which attempt to "place a value on human life"), the court held that Kurt had a right to pursue his claim. The Saskatchewan Court of Appeal denied the physician leave to appeal, and remanded the case back to trial to ensure a full hearing, rather than upholding the decision on a preliminary application to strike out the wrongful life claim. [416]

## 6.6 Disability Jurisprudence – Significant Cases of the Supreme Court of Canada – *Rodriguez, Latimer,* and *Eldridge*

The following cases, decided by the Supreme Court of Canada, begin to articulate important aspects of the legal understandings of, and protections afforded to, the disabled. They make clear the position of the Supreme Court regarding whether a second party may terminate the life of a (living) person with disabilities in the absence of criminal charges – it may not. It is also clear that the Supreme Court has placed an onus upon the provincial governments to alleviate the effects of programs and services (or the lack thereof) that perpetuate categorical discrimination against people with disabilities.

In *Rodriquez* v. *British Columbia (Attorney General)*,[417] Sue Rodriguez, who was suffering from ALS, applied to the court for an order that s. 241(b) of the *Criminal Code*, that prohibits the giving of assistance to commit suicide, be declared invalid on the ground that it violated her rights under ss. 7, 12 and 15(1) of the *Charter*. Sue Rodriguez wanted the ability to end her own life and to control the circumstances, timing and manner of her own death. The majority of the Supreme Court, in refusing to strike down the legislation, described s. 241(b) as, "fulfil[ling] the government's objective of protecting the vulnerable." This provision was found to be grounded in the state's interest in protecting life, and based on the principle of "sanctity of life" that holds that life should not be depreciated by allowing life to be taken. The British Columbia Court of Appeal, on the other hand, considered the case to revolve not around "life and death," but around the notion of "quality of life." On this basis, it was held that the lawful right of a terminally ill person to terminate her life was protected under the *Charter*. McEachern C.J held, "[i]n my view, death and the way we die is a part of life itself." The issue of human dignity was clearly framed as one of fundamental justice, and "any provision which imposes an indeterminate period of senseless physical and psychological suffering upon someone who is shortly to die anyway cannot conform with any principle of fundamental justice.[418] McEachern C.J. applied the Oakes test, and found that the violation of s. 7 by the operation of s. 241 of the *Code* could not be justified under section 1 of the *Charter*, as it did not impair the rights of the terminally ill, or of the appellant in particular. At the Court of Appeal, Section 241(b) was held to be inoperative as it applied to Ms. Rodriguez and her physician. She was permitted to proceed to arrange for physician-assisted suicide, provided certain conditions of competency, intention, and free will were met. The Supreme Court of Canada reversed this decision.

At the Supreme Court, Sopinka J., writing for the majority, held that no such deprivation of fundamental justice or liberty interest arose as a result of the *Criminal Code* provision. Security of the person, according to Sopinka J. pertains to a *living* person, and does not entail the ability to "choose," "or to manage" one's death. He stated, "death is, for all mortals, inevitable. Even when death appears imminent, seeking to control the manner and timing of one's death constitutes a conscious choice of death over life. It follows that life as a value is engaged even in the case of the terminally ill who seek to choose death over life."[419] By acknowledging that control over one's body falls squarely within the notion of security of the person, Sopinka J. found that the prohibition in s. 241(b) deprived Rodriguez of autonomy over her person, as it caused her physical pain and psychological stress. Given a legal context in which suicide itself is not unlawful, and the common law allows a physician to withhold or withdraw life-saving or life-maintaining treatment on the patient's instructions and to administer palliative care that has the effect of hastening death, the Court had to determine whether the existence of a criminal prohibition on assisting suicide for a person in the appellant's situation is contrary to principles of fundamental justice. Sopinka J. framed this inquiry as being "whether the blanket prohibition on assisted suicide is arbitrary or unfair in that it is unrelated to the state's interest in protecting the vulnerable, and that it lacks a foundation in the legal tradition and societal beliefs that are said to be represented by the prohibition."[420] Given the grave importance of preventing abuses in the context of assisted suicide, the court held that the prohibition was supported. "In upholding the respect for life, it may discourage those who consider that life is unbearable at a particular moment, or who perceive themselves to be a burden upon others, from committing suicide."[421] In separate reasons, the then Chief Justice Lamer, concluded that disabled persons who are unable to commit suicide without assistance are discriminated against contrary to s. 15 in that they are deprived of a benefit or subjected to a burden by virtue of s. 241(b) of the *Criminal Code*. However, he found the infringement to be justified under s.1 of the *Charter*.

In a dissenting opinion, McLaughlin J. chose to frame the case as one interrogating the right of the state to limit the right of a person to make decisions about her body. She opined that the denial of the ability to commit suicide to Sue Rodriguez – a choice available to others – could not be justified. McLaughlin relied upon the reasoning of the majority in *R.* v. *Morgentaler*[422] to demonstrate the way in which the provision denied Rodriguez the ability to make decisions about her body, which "affected only her body."[423] She forcefully argued that

> [t]here may be no reason on the facts of Sue Rodriguez's case for denying to her the choice to end her life, a choice that those physically able have available to them. Nevertheless, she must be denied that choice because of the danger that other people may wrongfully abuse

> the power they have over the weak and ill, and may end the lives of these persons against their consent. Thus, Sue Rodriguez is asked to bear the burden of the chance that other people in other situations may act criminally to kill others or improperly sway them to suicide. She is asked to serve as a scapegoat.[424]

Based on the principles of fairness, she concluded that the prohibition *was not* justified under s.1.

*R.* v. *Latimer* [425]deals with the issue of disability, in the context of whether a parent may end the life of a severely disabled child without penalty under the criminal law. Robert Latimer killed his 12-year-old daughter Tracy by carbon monoxide poisoning as a result of his belief that he must put an end to her suffering. Tracy had a severe form of Cerebral Palsy that rendered her quadriplegic, immobile, in chronic pain, and in need of constant care. In the words of the Supreme Court, Tracy had a "serious disability, but [was] not terminally ill." The Latimers did not want to subject their daughter to further surgeries, or to intravenous feeding, and medication. They perceived these acts to be mutilations. It was on this basis that Robert determined that his daughter's life "was not worth living." At trial, Robert Latimer, charged with first-degree murder, was convicted of second-degree murder.[426]

Upon appeal to the Supreme Court of Canada, Robert argued that the defence of necessity should have been available to him at trial. The defence of necessity entails the belief that "a liberal and humane criminal law cannot hold people to the strict obedience of laws in emergency situations where normal human instincts, whether of self-preservation or altruism, overwhelmingly impel disobedience." There are three requirements: an urgent situation of "clear and imminent peril"; no reasonable legal alternative to disobeying the law; and proportionality between the harm inflicted and the harm avoided. This test, applied on an objective standard, must convince the trial judge that the beliefs of the accused held an "air of reality." The Supreme Court, while recognizing the availability of the defence, ruled that none of the three requirements were fulfilled. In so doing, this case articulates the legal understanding of harm in the context of disability.

> The harm inflicted in this case was ending a life; that harm was immeasurably more serious than the pain resulting from Tracy's operation that Mr. Latimer sought to avoid. Killing a person – in order to relieve the suffering produced by a medically manageable physical or mental condition – is not a proportionate response to the harm represented by the non-life-threatening suffering resulting from that condition.[427]

The Court tempered its investigation of the defence with the need to "respect the life, dignity and equality of those affected by the act in question," and asserted that "the fact that the victim... was disabled... does not affect our conclusion." Just as the decision to deny the defence was upheld, so too was Robert's mandatory minimum sentence of ten years for second-degree murder on the basis of the extreme nature of the crime, the deliberation and planning required to commit the crime, and upon his obvious lack of remorse. Neither Robert's good character nor his love for his daughter were found to mitigate these factors.

In *Eldridge* v. *British Columbia (Attorney General),*[428] the court considered whether a provincial government's failure to provide funding for sign language interpreters for deaf persons when they receive medical services violates s. 15(1) of the *Canadian Charter of Rights and Freedoms.* Neither of the two health care mechanisms in operation in British Columbia paid for sign language interpreters for the deaf.[429] This case arose when these services were no longer provided free of charge by a private, non-profit agency. It was asserted that the failure to provide the service resulted in a lower standard of health care provision for the hearing impaired. At trial, the application for a declaration of the *Charter* violation was dismissed on the basis that sign language interpretation was considered "ancillary to medically required services." Tysoe

J. held that the *Charter* does not require governments to implement programs to assist disabled persons, and that the government was under no obligation to provide "the benefit" in the first instance. The British Columbia Court of Appeal held that the lack of publicly funded interpreters was not discriminatory, as the majority located the responsibility for funding and service decisions as resting with particular hospitals, not as being mandated within the statutes. Hospitals, in this sense, where not considered "government," and thus not within the jurisdiction of the *Charter*. The majority of the Court of Appeal also held that s.15(1) of the *Charter* was not violated by the *Medical and Health Care Services Act* because it provides free health services to all persons – regardless of whether they are able to hear. In dissent, however, Lambert J.A. reasoned that those who are unable to hear experience difficulties communicating with health care providers, and that sign interpretation is not an "ancillary service." He held that the *Medical and Health Care Services Act* was discriminatory in situations where individuals "seek to obtain medical services that require, for the discharge of the practitioner's professional obligations, effective communication between the practitioner and the patient, and where effective communication can only be achieved through the provision of translation services." However, Lambert J.A. found the infringement justified, as the statute does not guarantee the free provision of *all* services.

The Supreme Court addressed four issues: whether, and in what manner, the *Charter* applies to the decision not to provide sign language interpreters for the deaf as part of the publicly funded scheme for the provision of medical care; whether this decision constitutes a prima facie violation of s. 15(1) of the *Charter*; whether, having found such a violation, it is saved by s. 1; and, if not saved by s.1, the Court had to consider an appropriate remedy. The Court found that the actions of hospitals and the Medical Services Commission, who exercise discretion conferred by the legislation potentially infringes the *Charter* – not the legislation itself. The *Charter* was found to apply to both entities in so far as they act pursuant to the powers granted to them by the statutes.

Within the s. 15(1) analysis, the Court, recognizing that as deaf persons, the appellants belong to an enumerated group under s. 15(1), embarked upon a discussion of the historical consequences of marginalisation and discrimination against the disabled. The Court attempted to recognize the extent to which the disabled are perceived as abnormal or flawed, and that the hearing impaired have, in fact, been subject to these constructions. Deaf persons, it was found, have been subjected to unequal treatment largely because of barriers to communication. While, on its face, the medicare system in British Columbia was found to apply equally to the deaf and hearing populations, the appellants claimed that the lack of funding for sign language interpreters renders them unable to benefit from the legislation to the same extent as hearing persons. The Court characterized this claim as one of "adverse effects discrimination," and reiterated that this type of claim is recognized as falling within s.15(1). The Court held that the adverse effects suffered by deaf persons stemmed not from the imposition of a burden not faced by the mainstream population, but rather from a failure to ensure that they benefit equally from a service offered to everyone.

The holding of the Supreme Court differed from the opinions of the trial and Court of Appeal by *not* presupposing a categorical distinction between state-imposed burdens and benefits. The Court placed a burden on the government to ameliorate the disadvantage that it neither helped create or exacerbate. Finding that effective communication is an integral part of the provision of medical services, the Court held that in circumstances in which deaf patients cannot communicate effectively with their doctors without an interpreter, they do not receive the same level of medical care as hearing persons. When assessing whether this was a reasonable limit, the Court held that the government had "manifestly failed to demonstrate that it had a reasonable basis for concluding that a total denial of medical interpretation services for the deaf constituted a minimum impairment of their rights."[430] The Court held that the appropriate and just remedy is to grant a declaration that this failure is unconstitutional and to direct the government of British Columbia to administer the statutes in a manner consistent with the requirements of s. 15(1). The Court left the decision of how to implement this directive to the government to be brought into force within a reasonable period of time.

## NOTES

[151] Health Canada describes the international situation:

> In the United Kingdom, the Human Fertilisation and Embryology Authority has been the national government regulatory body in the field of reproductive and genetic technology since 1990. The Authority has the power to regulate and license research and clinical practice; develop national standards in the form of a Code of Practice; inspect and monitor compliance with the regulations; and maintain a national registry of treatments. France has implemented broad bioethics legislation, which includes issues of assisted reproduction, and restrictions on RGT practices through licensing and prohibitions. National advisory bodies on bioethics and RGT licensing have also been established. In Australia, most states and territories have professional standards and ethical guidelines; they regulate and license RGT research and clinical practice, prohibit surrogacy arrangements and unlicensed research and clinical practice, and have RGT advisory and licensing bodies with public input.
>
> Prior to developments in 2001, the United States did not have a national regulatory regime; some states regulate RGTs within their state health departments. Following international cloning developments, President Clinton called for a moratorium on the use of federal funds to support any attempt to create a child by somatic cell nuclear transfer (the technique used in the animal world to create Dolly, the sheep). On July 31, 2001 a bill was passed in the United States House which bans all human cloning. The cloning of people either to produce children, or to create embryos for medical research is now a federal crime, punishable by up to 10 years in prison and fines of up to $1 million (U.S.) or more. The House-passed ban does not affect cloning to produce molecules, DNA, cells other than human embryos, tissues, plants or animals. The bill is now before the Senate.

See Health Canada, Reproductive and Genetic Technologies: Overview Paper (1999) *http://www.hc-sc.gc.ca/english/protection/biologics_genetics/reproduction/rgt/overview.htm* .

[152] Caulfield (1995), p. 313.

[153] The Royal Commission on New Reproductive Technologies (1993), 767-768.

[154] Sidarous & Lamothe (1995), p. 153.

[155] For example, Bartha Knoppers refers to a recent law in Norway on assisted reproduction and genetics which mandates comprehensive genetic counselling for genetic tests required for pre-symptomatic diagnosis, predictive diagnosis and for the detection of carriers of hereditary diseases. Genetic tests for general diagnostic purposes, however, are exempt from mandatory counselling. See, Knoppers (1994), pp. 8-11. She notes that other bodies recommend that full disclosure cover unexpected findings (from testing to research), which are to be communicated if they are of direct clinical importance to the person or his/her family. See, Health Council of the Netherlands (1994) which states at 73 that any information derived from stored tissue that eventually becomes relevant should be communicated only if the individual concerned has explicitly requested this in advance. Knoppers (1995), pp. 11-12.

[156] Mykitiuk & Wallrap (1999), p. 351.

[157] For a discussion of how genetic counselling services are affected by these conditions, see Williams-Jones (1999), pp. 49-67.

[158] For an outline of these practices, see, *New Reproductive and Genetic Technologies: Setting Boundaries, Enhancing Health* (Health Canada, June 1996). An Advisory Committee on the Interim Moratorium was created in January 1996 to monitor compliance with the moratorium, and to advise the government on follow-up action. In addition, the Committee follows the development of new reproductive and genetic technologies, and identifies emerging practices that should be included in the moratorium. The moratorium remains in effect until the prohibitions become law.

[159] Mykitiuk & Wallrap (forthcoming 2002).

[160] Canadian Medical Association (1997), p. 7.

[161] Canadian Medical Association (1996).

[162] Ibid., *supra* note 10, pp. 7-8.

[163] As well, a process for public participation was implemented. The Committee scheduled hearings at various locations across the country. Ordinarily, a bill is introduced to the House of Commons rather than having a Standing Committee review draft legislation.

[164] Mykitiuk & Wallrap, *supra* note 9.

[165] Ibid.

[166] See sections 4. (5) and 7. (1), (2) of the Draft AHRA.

[167] Anyone convicted on indictment of a prohibition may face a fine of up to $500,000, or a jail term of no more than 10 years, or both. Summary convictions may result in a fine of up to $25,000, or a jail term of no more than 4 years, or both.

[168] The classes of controlled activities are not fixed however, as the Governor in Council may make regulations, "designating classes of controlled activities that may be authorized by a license issued under subsection 12(1)" of the draft AHRA. Section 40(1)(a). This section also refers to "classes of controlled activities that may not be authorized."

[169] Section 12.

[170] Health Canada (2001).

[171] Section 21. (2).

[172] Section 22.

[173] Section 19.

[174] Section 20.

[175] Section 22.

[176] Health Canada, *Reproductive and Genetic Technologies: Overview Paper, supra* note 1.

[177] Parliamentary Standing Committee on Health (2001).

[178] Ibid.

[179] Ibid., Recommendation 3(a), (e).

[180] Ibid., Recommendation 3(b), (c).

[181] Ibid., Recommendation 3(h).

[182] Ibid. Recommendation 3(d). The notion of the "integrity" of the human genome can be interpreted in two ways. The first, "integrity" as in wholeness, or completeness, or even "perfection" does not value the lives of persons with disabilities. However, "integrity" as it refers to the naturally occurring diversity of the human genome does recognize the inherent value of persons with disabilities.

[183] Ibid., Recommendation 9.

[184] Ibid., Recommendation 9, clause (3)(1)(h).

[185] Royal Commission on New Reproductive Technologies (1993), *supra* note 3, p. 79.

[186] Ibid., p. 801.

[187] Ibid., pp. 788-789.

[188] Caulfield (1995), *supra* note 2, p. 316. For a discussion of ownership and the patenting of genetic materials, see Gold (2000), pp. 413-435.

[189] See, Taylor & Mykitiuk (2001), pp. 65-71.

[190] Caulfield (2001), p. 17.

[191] Jamison (2001). The "balancing" referred to involves the attempt to find a middle ground between utopian and alarmist positions held regarding genetic technologies.

[192] Ibid.

[193] Ibid.

[194] Ibid.

[195] Ibid.

[196] McNamara & Nelson (1999), p. 53.

[197] Ibid., pp. 53-54.

[198] Ibid., p. 55.

[199] Ibid., pp. 56-59.

[200] Ibid., p. 69.

[201] Ibid.

[202] Ibid., p. 68.

[203] Sharpe (1996), p. 123.

[204] Caulfield (1995), *supra* note 2, p. 313. On this basis, Caulfield questions whether uniform guidelines are possible, necessary, or desirable.

[205] Royal College of Physicians and Surgeons of Canada (2000), p. 5.

[206] Ibid., p. 3.

[207] Ibid.

[208] Ibid., p. 4.

[209] Ibid.

[210] Ibid.

[211] Canadian College of Medical Geneticists (1993).

[212] Canadian Association of Genetic Counsellors (1999), p. 7.

[213] Ibid.

[214] Ibid.

[215] Ibid., p. 8.

[216] Ibid.

[217] Ibid., p. 15.

[218] Ibid.

[219] Society of Obstetricians and Gynaecologists of Canada (1998). These guidelines act in concert with those of the Canadian College of Geneticists, whose 1991 statement outlines the objectives of prenatal diagnosis,

1. To offer the widest possible range of informed choices to women at risk of having children with genetic abnormality;
2. To provide reassurance and reduce the level of anxiety;
3. To confirm the absence of disease in question in high-risk women in order to allow them to continue a pregnancy; and
4. To facilitate optimal treatment of affected infants through early diagnosis.

See, Ethics and Public Policy Committee, Canadian College of Medical Geneticists (1991), p. 1129.

[220] Society of Obstetricians and Gynaecologists of Canada (1993).

[221] Ibid. [emphasis added].

[222] Society of Obstetricians and Gynaecologists of Canada (1998), p. 3.

[223] Ibid., p. 3.

[224] Ibid., p. 3.

[225] Ibid., p. 4.

[226] Ibid., p. 4.

[227] Ibid., p. 4 [emphasis added].

[228] Ibid., p. 4.

[229] NSERC (1999).

[230] This term is neither defined nor expanded upon.

[231] NSERC (1999), *supra* note 79.

[232] Ibid., Article 8.5.

[233] Ibid.

[234] Ibid., Article 8.1.

[235] Ibid., Article 8.2.

[236] Ibid., Article 8.3.

[237] Ibid.

[238] Ibid.

[239] Ibid., Article 8.4.

[240] Ibid.

[241] Ibid.

[242] Ibid.

[243] Ibid.

[244] Dossetor & Briggs (1995), p. 297.

[245] Burgess, Laberge & Knoppers (1998), p. 1310.

[246] Caulfield (1995), *supra* note 2, pp. 308-309.

[247] Ibid., pp. 312-313.

[248] Ibid., p. 313.

[249] Fraser (1995), pp. 304-305.

[250] For a more detailed summary of the history of law pertaining to abortion in Canada, see, Rodgers (1999), pp. 276-287.

[251] *Morgentaler* v. *Queen* (1988), 82 N.R. 1.

[252] [1989] O.J. No. 1587.

[253] [1989] 2 S.C.R.

[254] [1996] B.C.J. No. 3001.

[255] S.B.C. 1995, c. 35, ss. 2(1), 2(1)(a), 2(1)(b), 3, 4, 5, 6.

[256] [1997] 3 S.C.R. 925. This case is discussed in more detail in the section entitled, "Prenatal Liability."

[257] For a summary of the arguments in favour of foetal rights, see, Shaffer (1994).

[258] 112 C.C.C. (3d) 481.

[259] (1991), 63 D.L.R. (3d) 97. This case arose from charges against two midwives after the death of an unborn child while it was in the birth canal. In the Supreme Court decision, at page 106, Chief Justice Lamer said that it was clear from the wording of Section 206 that a foetus is not a "human being" for the purposes of the *Criminal Code*. The two midwives were acquitted of both the offence of criminal negligence and criminal negligence causing death and that decision was upheld by the Supreme Court of Canada.

[260] This was articulated by the Ontario Court (General Division) in *Drummond* as follows,

> Save for Subsection 238(1), which deals with killing an unborn child in the act of birth, Canada has set no time frame to fix a point in time, following conception, after which the killing of a foetus is only allowed in limited circumstances, such as, for example, to save the life of the mother.

[261] *Abortion Act*, 1967, s. 1(1)(d). Three Australian states have modeled similar provisions on the UK legislation: South Australia, the Northern Territory, and Western Australia. Abortions can be legally performed in these jurisdictions if the risk to the pregnant woman's life or to her physical or mental health is greater than it would be if the pregnancy were not terminated and in cases where a substantial risk exists that the child will be born with a physical or intellectual disability. The abortion must be approved by two medical practitioners and must be performed in a hospital. In addition, in the Northern Territory, it must be performed during the first 14 weeks of pregnancy, except in the case of a threat to life or physical or mental health, or a risk of serious injury. Several states in the United States also have similar legislation.

[262] Disability Rights Commission, "DRC Position on Section 1(1)(d) of the Abortion Act 1967" http://www.drc-gb.org/drc/InformationAndLegislation/Page322.asp .

[263] Sharpe (1996), *supra* note 53, p. 123.

[264] Ibid., p. 123.

[265] Knoppers (1999), pp. 365-366.

[266] Ibid., 365.

[267] *H(R)* v. *Hunter* (1996), 32 CCLT (2d) 44 (Ont. Ct. [Gen. Div.]); *Arndt* v. *Smith* (1997), 148 DLR (4th) 48 (SCC); For further discussion, see, Caulfield & Robertson (1996), pp. 59-79.

[268] Picard & Robertson (1996) in Caulfield (2001), pp. 143-147.

[269] Sidarous & Lamothe, *supra* note 4, pp. 155-56. Sidarous and Lamothe note at 156 (note 8), however, that as a response to shortages of specialized genetic counsellors, primary care providers can "perform counselling for specific disorders within a screening program." For a more detailed investigation of available support groups, *see* the Canadian Directory of Genetic Support Groups at http://www.lhsc.on.ca/programs/medgenet/support.htm .

[270] Bartha Knoppers (1999), *supra* note 115, p. 366.

[271] Burgess, et al. (1998), *supra* note 95, p. 312.

[272] Knoppers (1999), *supra* note 115, p. 366.

[273] Ibid. There are also concerns that general practitioners who do counsel their patients may not have sufficient training. See, Burgess, et al. (1998), *supra* note 95.

[274] Sharpe (1996), *supra* note 53, p. 124.

[275] Sharpe (1997), pp. 397-398.

[276] Sharpe (1996), *supra* note 53.

[277] Ibid.

[278] Caulfield, (1995), *supra* note 2, p. 315. See, *Ciarlarliello* v. *Schacter*, [1993] 2 S.C.R. 119; *Adan* v. *Davis*, [1998] O.J. No. 3030 (QL) (Gen. Div.).

[279] Sharpe (1997), *supra* note 125, p. 389. Sharpe points to the professional guidelines of the RCPS and the CCMG to support his point. See, "Policy/Guidelines."

[280] Ibid., p. 420.

[281] Ibid., p. 423.

[282] Ibid., p. 430.

[283] Ibid.

[284] Ibid., pp. 428-430 [emphasis added].

[285] Ibid., pp. 439-440.

[286] Ibid., p. 441.

[287] Ibid., p. 442.

[288] In 1983, the President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research made recommendations for the disclosure of genetic data which have been widely adopted by subsequent commentators in Canada and the United States. *See, President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research, Screening and Counselling for Genetic Conditions* (Washington, D.C.: The President's Commission, 1983). The Commission recommended that genetic data should be disclosed to biological relatives without patient consent, only under the following circumstances:

> 1. reasonable attempts to elicit voluntary disclosure are unsuccessful; 2. there is a high probability of serious (e.g. irreversible or fatal) harm to an identifiable relative; 3. there is reason to believe that disclosure of the information will prevent harm to the relative; and 4. the disclosure is limited to the information necessary for diagnosis or treatment of the relative.

These guidelines were adopted in the U.S. by the Committee on Assessing Genetic Risks, Division of Health Sciences Policy, Institute of Medicine, in its 1994 report; *Assessing Genetic Risks*, supra note 39 at 278, and the Science Council of Canada, *Genetics in Health Care: Report 42* (Ottawa: Minister of Supply and Services Canada, 1992). Flanagan, at 281, offers further insight into the important, yet vague terms employed within the guidelines,

> In stating that disclosure is only justified if there is a "high probability" that the relative is affected by a "serious" genetic disorder, the guidelines suggest that the relative must bear a significantly higher risk than the general population of being affected by an "irreversible or fatal" genetic disorder. The information may be of even greater significance if it is unlikely that the defect will be detected without disclosure. The guidelines also suggest that disclosure should enable the "prevention of harm," for example, by enabling the affected relative to seek available treatment (or avoid harmful stimuli).

See, Flanagan (1995), pp. 281-282.

[289] Knoppers (1999), *supra* note 115, p. 357.

[290] Knoppers, however, asserts that there may be an ethical duty to do so. She points to the existence of American ethical guidelines on this point. See, American Society of Human Genetics, ASHG Statement (19980, p. 474. Knoppers (1999), *supra* note 115, p. 366 and note 54.

[291] Ibid., p. 362.

[292] *Furniss* v. *Fitchett*, [1958] N.Z.L.R. 396; *Peters-Brown* v. *Regina District Health Board*, [1996] 1 W.W.R. 337 (Sask. Q.B.), aff'd [1997] 1 W.W.R. 639 (C.A.).

[293] Flanagan (1995), *supra* note 138, p. 269. For a detailed account of the laws of medical confidentiality, see, Oscapella (1993), p. 172.

[294] Commentators note, however, that "there remains a marked absence of extensive legislative intervention in the area of medical confidentiality." See, Flanagan (1995), *supra* note 138.

[295] Flanagan (1995), *supra* note 138, p. 274.

[296] *McInerney* v. *MacDonald*, [1992] 2 S.C.R. 138; 93 D.L.R. (4th) 415; *Norberg* v. *Wynrib* (1992) 92 D.L.R. (4th) 449 (S.C.C.); *Henderson* v. *Johnston* (1956) 5 D.L.R. (2d) 524 (Ont. High Ct.); *Cox* v. *College of Optometrists of Ontario* (1988) 65 O.R. 461 (Ont. High Ct.).

[297] Barbara von Tigerstrom, Mylène Deschênes, Bartha Maria Knoppers and Timothy A. Caulfield articulate how ethical codes influence the duty of confidentiality. They state,

> Health professionals also have ethical duties of confidentiality contained in, for example, the Hippocratic Oath and the codes of ethics of various professional associations. Although these are not legal duties per se they have some indirect force in law because a health professional who breaches them may be subject to discipline by a professional body under the authority of provincial legislation. These ethical duties may also be referred to as standards of conduct for the profession and thus be relevant to, for example, an action in negligence. Notwithstanding such ethical duties, a health professional is permitted to disclose information where disclosure is required by law.

Von Tigerstrom, et al. (2000), p. 12.

[298] Ibid., p. 12. Although there is not yet Canadian case law on point, they refer to the American case of, *Herdrich* v. *Pegram*, 1998 LEXIS 20189 (7th Cir. August 18, 1998) .

[299] Flanagan (1995), *supra* note 138, p. 276 [emphasis added].

[300] (1992), 93 D.L.R. (4th) 415 (S.C.C.).

[301] *McInerney*, *supra* note 146, pp. 427-428.

[302] Flanagan (1995), *supra* note 138, p. 279. *See, Wellesley Hospital* v. *Lawson* (1977), 76 D.L.R. (3d) 688 (S.C.C.); *Tanner* v. *Norys* [1980] 4 W.W.R. 33 (Alta. C.A.); *Wenden* v. *Trikha* (1991), 116 A.R. 81; aff'd (1993), 14 C.C.L.T. (2d) 225 (Alta. C.A.); leave to appeal to the S.C.C. refused (1993), 17 C.C.L.T. (2d) 285n.; *Pittman Estate* v. *Bain* (1994), 112 D.L.R. (4th) 257 (Ont. Ct. Gen. Div.)(currently under appeal). These cases all dealt with psychiatric patients who presented a "reasonably foreseeable" risk of violent assault or reckless behaviour that could seriously endanger the safety of others. Flanagan notes

that it will be difficult to determine precisely those to whom the duty to warn would be owed, and the types of risk that might give rise to the duty.

[303] Nelson, Haymond & Sidarous (1998), pp. 106-107.

[304] *Eldridge* v. *British Columbia (Attorney General)*, [1997] 3 S.C.R. 624, at 676-77.

[305] Dickens (1999), p. 117.

[306] *Malette* v. *Shulman* (1990), 67 D.L.R. (4th) 321 (Ont. C.A.).

[307] Dickens (1999), *supra* note 155, p. 117.

[308] See, *Fleming* v. *Reid* (1991), 82 D.L.R. (4th) 298 (Ont. C.A.) It was also reaffirmed in *Nancy B.* v. *Hotel-Dieu de Quebec* (1992), 86 D.L.R. (4th) 385 (Que. Sup. Ct.).

[309] Robertson, et al. (1995), p. 191. The three other requirements of a valid consent are: the person consenting must have capacity to do so; the consent must be specific to the treatment and to the person who is to administer the treatment; and the consent must be informed.

[310] Ibid., p. 191.

[311] Dickens (1999), *supra* note 155, p. 125.

[312] [1980] 2 S.C.R. 192.

[313] von Tigerstrom, et al. (2000), *supra* note 167. Dickens (1999), *supra* note 155, reminds us that the question of whether a particular risk is a material risk is a matter for the trier of fact. It is also for the trier of fact to determine whether there has been a breach of the duty of disclosure. See, *Videto* v. *Kennedy* (1981), 125 D.L.R. (3d) 127 (Ont. C.A.).

[314] Knoppers (1999), *supra* note 115, p. 366.

[315] Ibid.

[316] Caulfield (1995), *supra* note 2, p. 315.

[317] (1990), 72 O.R. (2d) 417 (Ont C.A.).

[318] [1993] 2 S.C.R. 119.

[319] Knoppers (1999), *supra* note 115, p. 367.

[320] Ibid. See, *B.(R.)* v. *Children's Aid Society of Metropolitan Toronto*, [1995] 1 S.C.R. 315.

[321] Ibid.

[322] Caulfield (1995), *supra* note 2, p. 316.

[323] Sharpe (1997), *supra* note 125, p. 430 [emphasis added].

[324] Ibid., pp. 433-434 [emphasis added].

[325] Robertson (1999, p. 92. See, *Snell* v. *Farrell* (1990), 72 D.L.R. (4th) 289 (S.C.C.).

[326] Sharpe (1997), *supra* note 125, p. 434.

[327] *Snell, supra* note 175.

[328] Robertson (1999), *supra* note 175, p. 93.

[329] Ibid.

[330] Crow (1998), p. 3 [emphasis added]. Crow asserts that rather than construing the non-disclosure of a material risk as a "harm," it should be understood as an "infringement of basic human dignity."

[331] Sharpe (1997), *supra* note 125, p. 434.

[332] Ibid.

[333] Ibid., pp. 435-436.

[334] Ibid., p. 450.

[335] Ibid., pp. 450-451.

[336] Ibid., p. 450 note 229.

[337] Robertson (1999), *supra* note 175, p. 83.

[338] Robertson, et al. (1995), *supra* note 159, pp. 210-211.

[339] Robertson (1999), *supra* note 175, p. 93.

[340] [1933] 4 D.L.R. 337.

[341] (1972), 26 D.L.R. (3d) 418 (Ont. H.C.).

[342] [1932] A.C. 562.

[343] The court however, explicitly refrained from any discussion of the legal rights of a child *en ventre sa mere* or of a foetus.

[344] This has been codified in the Ontario *Family Law Act*, R.S.O. 1990, c. F-3, s. 66, which states that "[n]o person is disentitled from recovering damages in respect of injuries for the reason only that the injuries were incurred before his or her birth."

[345] "Mother" in this context (among others) is not without controversy. For example, a timely question in light of the practice of surrogacy is whether a tort action lie against a gestational surrogate if her actions during her pregnancy result in an injury to the child. This raises important questions about the legal understanding of "mother." Chisick and Baccus highlight some of the elements of this debate:

> ... potentially tortious behaviour is more likely on the part of a gestational surrogate than a natural mother because of the competing interests at play in the former situation; although the priorities of a natural mother (and, it should be observed, of intended parents) might be expected to rest squarely with the well-being of the foetus, the surrogate mother might well place her own health or well-being first. Where this type of maternal-foetal conflict exists, it is quite conceivable that the surrogate might act without the knowledge or approval of the intended parents. Where these actions result in injury to the child, the intended parents may be inclined to sue the surrogate, whether to help defray the costs of caring for the injured child, to compensate the child for his or her loss, or even to attempt to punish the surrogate mother for her misconduct.

They note that to justify an award of punitive damages, the surrogate's actions would likely need to have been intentional and of a high-handed, oppressive or malicious character. See, *Hill* v. *Church of Scientology*, [1995] 2 S.C.R. 1130. See generally, Chisick & Baccus (1997), pp. 49-75.

[346] (1996), 138 D.L.R. (4th) 254 (Man. C.A.), rev'ng (1996), 138 D.L.R. (4th) 238 (Q.B.) Hereafter, "*(G.)(D.F.)*."

[347] RCNRT, *supra* note 3, pp. 964-965.

[348] Prior to this case, state action on behalf of the foetus was found to be justified to protect it from the pernicious actions of the women. Such action relied on the application of provincial child protection and mental health legislation. See, *Re Children's Aid Society of City of Belleville, Hastings County and T et al.* (1987), 59 O.R. (2d) 204 (Fam. Ct.); *Re Baby R.*, (1988), 53 D.L.R. (4th). Intervention was not permitted on the basis of the objections of a third party, however, in *Tremblay* v. *Daigle* (1989), 62 D.L.R. (4th) 634. For a complete discussion of the cases preceding *(G.)(D.F.)*, see, Rodgers (1993), pp. 33-46.

[349] The common law aversion to investigations into "alleged parental indiscretions during pregnancy, like excessive smoking, drinking, or taking drugs," was reiterated. The reasons for this aversion are twofold - due to respect for the mother's rights, and on the basis of the concern that the mother may grow to resent her child if forced to deviate from her chosen course of behaviour. See also *Re F (in utero)*, [1988] 2 All E.R. 193 (C.A.), a case with similar facts and holdings to those in the *G.(D.F.)* case. The court in *Re F* also took the position that conflicts between the rights of the mother and the unborn child are "most undesirable."

[350] [1997] 3 S.C.R. 925.

[351] *G.(D.F.)*, p. 993.

[352] *G.(D.F.)*, p. 988. Here Major J. was relying on *Dobson (Litigation Guardian of)* v. *Dobson* (1997), 148 D.L.R. (4th) 332 (N.B.C.A.), at that time on appeal to the Supreme Court of Canada. This relies on an extension of the "foetus as legal fiction" argument, similar to that made in the "driving" cases, as well as in wrongful life claims (see "Wrongful Birth/Wrongful Life").

[353] [1999] 2 S.C.R. 753 (S.C.C.).

[354] [1997] N.B.J. No. 17.

[355] [1997] N.B.J. No. 232 (N.B.C.A.).

[356] Robertson, et al. (1995), *supra* note 159, p. 188.

[357] Caulfield & Robertson (1996), p. 77.

[358] Gerald Robertson, et al. (1995), *supra* note 159, p. 201.

[359] Dworkin (1996), p. 90.

[360] Ibid., p. 86.

[361] Ibid.

[362] Nelson & Robertson (2001), p. 104.

[363] Ibid., p. 104. For a discussion of the low success rates for plaintiffs in informed consent cases, see Robertson (1991), p. 423. Also, Timothy Caulfield warns of the possibility of misuse of tests due to fear of physician liability. At 16, he states,

> ... law may also exacerbate a number of the commercialization problems by encouraging physicians to utilize genetic services. As testing becomes more common and as patients' expectations rise, physicians may feel the need to practice defensive medicine. That is, physicians may succumb to the demands of patients or may believe that they should simply recommend testing in order to avoid liability. Because of the high profile nature of wrongful birth law suits, defensive practices might be particularly problematic in relation to the use of prenatal diagnosis. Given the invasive nature of genetic testing and PND in particular, physicians should strive to find more constructive ways to address liability concerns (e.g., enhancing communication skills).

See, Caulfield (2001), *supra* note 40.

[364] Dworkin (1996), *supra* note 209, p. 86. Dworkin, at 93, notes the procedural difficulties in establishing that the defendant's negligence caused the harm. This is due to the fact that, "there is seldom any evidence other than the parent's testimony about what they would have done if they had been properly informed."

[365] Ibid., at 86.

[366] Caulfield (2001), "Liability", p. 145.

[367] Nelson & Robertson (2001), *supra* note 212, p. 103.

[368] In fact, questions about the nature of the physician/counsellor's duties continue to emerge. Sidarous and Lamothe, *supra* note 4, p. 169 ask, does a genetic counsellor have an obligation to inform patients of testing or therapies that become available after the consultation has been performed?

[369] (1978), 86 D.L.R. (3d) 719 (Ont. H.C).

[370] (1979), 3 Med. L.Q. 72 (Alta. T.D.).

[371] The amount of the damages claimed was the cost of raising the one-year-old child, until she reached the age of 21.

[372] (1987), 114 D.L.R. (3d) 585 (Que. Sup. Ct.).

[373] Ibid.

[374] (1984), 48 Nfld. & P.E.I. 102 (Nfld. T.D.).

[375] Robertson, et al. (1995), *supra* note 159, p. 202. Robertson et al. remark upon the odd nature of this case. The possibility of "mitigation by adoption" has not arisen since.

[376] *Arndt* v. *Smith* (1997), 148 D.L.R. (4th) 48 (S.C.C.).

[377] Crow, (1998), *supra* note 180, p. 3.

[378] (1997), 148 D.L.R. (4th) 48 (S.C.C.).

[379] [1998] O.J. No. 4683 (Ont. Gen. Div.) In this case, the court also relied upon the limited severity of the child's disabilities, the quality and stability of Mrs. Mickle's marriage, and the limited risks to her health to determine that she would not have had an abortion had she been apprised of the risks withheld as a result of a faulty ultrasound procedure.

[380] *Arndt, supra* note 226, p. 83.

[381] Ibid., p. 83.

[382] Nelson & Robertson (2001), *supra* note 212, p. 103 [emphasis added].

[383] [1996] O.J. No. 4477.

[384] (1997), 154 D.L.R. (4th) 707 (B.C.S.C.).

[385] Ibid.

[386] These categories were then reiterated in *Jones (Guardian ad litem of)* v. *Rostvig* [1999] B.C.J. No. 647 (S.C.) (QL), where the Supreme Court of British Columbia explicitly framed the issue of a child born with Down Syndrome after the failure of his mother's physician to provide genetic testing as an investigation of whether there had been a breach of duty toward the mother (i.e. for loss of opportunity to have an abortion), rather than toward the child (i.e. for pecuniary damages resulting from "wrongful life"). On potential future causes of action arising in the genetic context, Pattison states at 20

> Parents might also wish to bring a "wrongful birth action" alleging a negligent failure to prevent the conception or birth of a child with a particular trait (e.g., a congenital defect), or a negligent failure to ensure the birth of a child with a particular trait. The... situation... exists when, for example, there is a negligent failure to advise the parents of the risk of having a child with a congenital abnormality. Other examples include a negligent failure to carry out a prenatal diagnostic technique (at all or with due care) when this would have disclosed a congenital defect. In the future, this type of action could arise as a result of undesired consequences following from negligent attempts at conducting germ-line gene therapy, or cloning.

See, Pattison (1999), pp. 19-32.

[387] R.S.B.C. 1996, c. 128, ss. 87 [am. S.B.C. 1997, c. 20, s. 16], 88(1) [idem, s. 17], 91(3).

[388] (2000), 184 D.L.R. (4th) 251.

[389] [2002] S.C.J. No. 8.

[390] [1999] B.C.J. No. 650 (B.C.S.C.).

[391] Robertson, et al. (1995), *supra* note 159, p. 202; Caulfield (2001), *supra* note 213, p. 144. See, *Hunter, supra* note 241.

[392] Nelson & Robertson (2001), *supra* note 212, p. 104. See, *McFarlane* v. *Tayside Health Board* [1999] 4 All E.R. 961 (H.L.).

[393] This argument is made by Nelson and Robertson, *supra* note 212, notwithstanding two Canadian cases in which damages were awarded to parents of a "healthy" child in wrongful birth cases. See, *Suite* v. *Cooke* (1993), 15 C.C.L.T. (2d) 15 (Que. Supp. Ct.); aff'd [1995] R.J.Q. 2765 (C.A.); *Joshi* v. *Woolley* (1995), 4 B.C.L.R. (3d) 208 (S.C.).

[394] The action is usually brought by a legal guardian, on behalf of the child.

[395] Pattison (1999), *supra* note 236, p. 21.

[396] Robertson, et al. (1995), *supra* note 159, p. 202.

[397] Pattison (1999), *supra* note 236, p. 21.

[398] Ibid., pp. 31-32.

[399] Nelson & Robertson (2001), *supra* note 212, p. 104.

[400] Pattison (1999), *supra* note 236, pp. 23-24.

[401] Harris (1998) in Pattison (1999), *supra* note 236, p. 24.

[402] Pattison (1999), *supra* note 236, p. 26. Based upon the cases of *Montreal Tramways* v. *Leveille* [1933] S.C.R. 456 and *Duval* v. *Sequin* (1973), 1 O.R. (2d) 482 (C.A.), Caulfield has argued that in Canada, "physicians providing information about genetic risk or advice on prenatal procedures can be viewed as owing a duty of care to the unborn child." See, Caulfield (2001), *supra* note 213, p. 144.

[403] One major hurdle, as recognized by Pattison (1999), *supra* note 236, p. 27, would be to establish that the defendant physician had the sole obligation to provide the plaintiff child with a monetary award.

[404] (1978) 114 D.L.R. (3d) 585 (Que. S.C.).

[405] [1990] B.C.J. No. 2576.

[406] [1992] B.C.J. No. 1687.

[407] The court contrasted the situation in this case to those in previous, unsuccessful wrongful life suits in which the child was not successful since a pre-existing condition – rather than the actions of the doctor – actually caused the injury sustained by the child after birth. See, *McKay* v. *Essex* [1982] 2 All E.R. 771.

[408] There is a body of American scholarship that argues in favour of wrongful birth actions. See, Strasser, (1999), pp. 29-75; Shapiro (1998), pp. 369-375; Shiffrin (1999), pp. 117-148; Jackson (1996), pp. 349-381. Similar controversy exists in Great Britain. For an analysis of both the jurisprudential and scholarly positions in England, see Pattison (1999), *supra* note 236.

[409] (1997), 148 D.L.R. (4th) 48 (S.C.C).

[410] England's leading case on the issue is *McKay* v. *Essex Area Health Authority*, [1982] 2 All E.R. 771 at 779, in which the English Court of Appeal held that a woman who contracted rubella while pregnant, which went undetected in a blood test by the local health authority, could claim damages on her own behalf for a lost opportunity of abortion, but the child could not claim a right to be "aborted or killed." In the United States case of *Becker* v. *Schwartz*, 386 N.E. 2d 807 (1978), the New York Court of Appeal held that a couple who were not advised of the availability of amniocentesis to test their foetus for Down Syndrome could not make a claim on their child's behalf for wrongful life. Some suggest that this case was decided without consideration of the "true" result of *Cherry* v. *Borsman*, thus creating uncertainty.

[411] [1998] O.J. No. 4683 (Gen. Div).

[412] [1999] M.J. No. 397 (Man. Q.B.).

[413] [1999] B.C.J. No. 647 (B.C.S.C).

[414] [1999] B.C.J. No. 650 (B.C.S.C.) (QL).

[415] [1998] S.J. No. 559.

[416] [1998] S.J. No. 645 (C.A.).

[417] [1993] 3 S.C.R. 519.

[418] Ibid., 538.

[419] Ibid., 587.

[420] Ibid., p. 596.

[421] Ibid., p. 540.

[422] [1988] 1 S.C.R. 30.

[423] *Rodriguez, supra* note 267, p. 618.

[424] *Rodriguez*, p. 621.

[425] [2001] 1 S.C.R. 3.

[426] [1997] A.J. No. 406. The Saskatchewan Court of Appeal upheld this conviction. [1998] S.J. No. 731.[427] *Latimer, supra* note 275, p. 27 [emphasis added].

[428] [1997] 3 S.C.R. 624.

[429] *Hospital Insurance Act*, R.S.B.C. 1979, c. 180 (now R.S.B.C. 1996, c. 204) and the *Medical and Health Care Services Act*, S.B.C. 1992, c. 76 (now known as the *Medicare Protection Act*, R.S.B.C. 1996, c. 286).

[430] *Eldridge, supra* note 278.

# 7

# LITERATURE REVIEW

## 7.1 Introduction

A review of scholarly literature addressing the issues of genetic testing and counselling picks up many of the same themes addressed elsewhere in this research report. The literature describes how the professional discourse of genetic counselling has emerged as a form of medical-therapeutic expertise focused on the minimization of an increasing number of potential risks posed to the "integrity" of genetic material. The role of the genetic counsellor is to frame these risks for their clients in a "value-free" non-directive manner so that clients can make "informed choices" of whether or not to be screened or tested for "abnormalities" in genetic material and to terminate an affected foetus.

The literature questions the "value-free" nature of counselling given that in the framing of the risks, a biased body of knowledge that labels genetic differences and people with disabilities as "abnormal" is conveyed to clients. As well, by presenting only certain "facts" and excluding information about the eugenic nature of the prenatal testing, of the arbitrariness of the identification of some genes, and not others, as "abnormal," of the effect this has on societal values and on the value accorded to people with disabilities draws into question the neutral, non-directive stance genetic counsellors purport to take.

Genetic counselling also imparts to women an understanding that prenatal procedures are inherently good, that pregnancies can be classified, imply risks, and demand supervision. In so doing, a woman's body is transformed into a field of operation for medical, technocratic, and bureaucratic interventions. This loss of control over their bodies is coupled with the loss of their free choice. In the context of government sanctioned prenatal testing programs and a legal milieu in which doctors can be sued for "wrongful births," women are mandated by medical professionals to arbitrate "normalcy" and to decide whether their foetus is fit to live or not. As such, the practice of genetic counselling can be seen to be a function of eugenic socio-political goals. Often unwittingly, women are being drawn into the process through the choices they are mandated to make, compromising their well-being, and human rights.

Finally, the literature on human rights points out that in addition, the trend toward geneticization and the application of genetic research risks undermining choice and self-determination for persons with disabilities. It also risks the commodification of children, where they are regarded as consumer objects, subject to quality control, the products of conception.

## 7.2 The Construction of Risk

Over the last several decades, "risk" (and its cohorts, chance, and uncertainty) have become a central organizing principle in Western societies, one that governs individual actions and responses aimed at promoting personal health and well-being, and at socio-political agendas and policies intended to ensure social security and stability. Knowledge and understanding of potential risks shape innumerable aspects of contemporary subjectivity and social life, including personal and group identities, self-perceptions, experiences of embodiment, daily schedules and routines, as well as individual and institutional interests and goals.[431] Indeed, an entire apparatus of expert research, knowledge and advice has developed around the concept of risk. Risk analysis, risk identification, risk assessment, risk evaluation, risk communication, and risk management, for example, are major fields of research and practice used to measure and control risk in areas as far-ranging as medicine and public health, finance, the law, business, and industry.

Lupton has argued that at least six major categories of "risk" that currently predominate in the concerns of individuals and institutions can be identified: environmental risks, such as those posed by pollution, radiation, chemical floods, fires, and dangerous road conditions; lifestyle risks, such as those related to the consumption of certain foods and drugs, engagement in sexual activities, driving practices, stress, leisure, and so on; medical risks, such as those that involve medical interventions or treatment (e.g., drug therapy, surgery, childbirth, reproductive and genetic technologies, and diagnostic tests); interpersonal risks, such as those related to intimate relationships, social interactions, love, sexuality, gender roles, friendship, marriage, and parenting; economic risks, including those implicated in unemployment, borrowing money, investment, and bankruptcy; and criminal risks, such as those that emerge if one participates in or is the potential victim of illegal activities.[432] That these categories of risk are prominent at this point in the history of Western societies, Lupton points out, is indicative of the broader socio-cultural, political, and economic context in which they have been generated, and in which they have acquired meaning. For at this historical moment, the meaning of risk has shifted from that of a neutral mathematical probability to one of impending danger.[433]

The professional discourse of genetic counselling can be regarded as one form of medical-therapeutic expertise. It has emerged in response to knowledge produced about an increasing number of potential risks posed to the "integrity" of genetic material. Genetic counsellors administer and convey information about genetic testing to clients who must make choices on the basis of the presence, or possible presence of, or predisposition to, any of several hundred "abnormalities" in genetic material that can be identified or predicted by a variety of methods and tests. The most commonly known and publicly available forms of this technology are prenatal testing and screening, which are intended to variously detect "abnormalities" in the development or projected development of a given foetus or embryo. For despite the fact that prenatal testing and screening are not recommended as universal procedures to be applied in all pregnancies, such practices are increasingly institutionalized within standard protocols for prenatal care and maternal "risk-management."[434] Because of this standardization of genetic testing within regimes of prenatal care, and given the escalating availability of genetic testing in general, genetics has fast emerged as an integral component of health care systems and services.

It has been remarked that because the provision of genetic counselling is not uniform, it is not possible to accurately describe the practice and discourse of prenatal genetic counselling. Biesecker and Hamby have nevertheless articulated four models to represent the variation within this discourse.[435] The four models are: the minimalist model, the informational model, the informed consent model, and the psycho-educational model. The authors explain these modes of counselling in the following ways:

### 1. The minimalist model

This form of counselling proceeds as a conversation between the provider and the client that occurs before routine prenatal testing (amniocentesis or CVS) for women over 35 years of age and those otherwise at risk of carrying a foetus with a condition for which a test is available. Counselling that proceeds according to this model emphasizes the risks of the test procedure itself rather than offering information about the foetus. Minimalist counselling does little to educate a client or prepare her for decision-making.

### 2. The informational model

A somewhat expanded model of counselling, the informational model involves discussion of chromosomes and non-disjunction as a cause of Trisomy. Studies have not consistently shown that this introduction to genetics assists clients in adjudicating the options available to them. Although this model assumes the benefit of such information, it neglects to consider client interpretations and significance of the information that does not resemble the providers' own priorities.

### 3. The informed consent model

This model of counselling assumes that a given client requires information about potentially identifiable "abnormalities" in the foetus or embryo, as well as information about the range of options and resources available regarding prenatal testing. Counselling that proceeds on the basis of this model involves more extensive discussion of the risks and benefits of testing than do the minimalist or informational models; in addition, it strives to uncover the personal meaning of the information conveyed. This model of counselling is deliberative; a dialogue between the client and the counsellor. Although genetic counsellors are trained to provide this type of decision-making counselling, time restraints, conflicting agendas with obstetricians and physicians, and limited reimbursement for such time consuming services restrict its availability.

### 4. The psycho-educational model

This is the most extensive model of counselling, for it includes information about the conditions and impairments under examination; in addition, it involves discussion of client interpretations and perceptions of the information. The psycho-educational approach proceeds as a collaborative endeavour in which the counsellor attempts to achieve an empathetic understanding of the client's values with respect to childbirth, terminating a pregnancy, and establishing a family. Within this model, furthermore, a skilled counsellor can establish a sufficiently therapeutic relationship with a pregnant woman in order to probe the reactions of that client to the possibility that an "abnormality" in the foetus will be detected. In addition, the client's experiences with people with disabilities may be explored in the context of this approach to counselling. This model requires a great deal of skill and flexibility on the part of the counsellor. It also takes a significant amount of time and is often difficult to implement, for it requires that a given counsellor be very self-aware and insightful about her or his own beliefs and values with respect to childbirth, disability, and abortion.

While Biesecker and Hamby's models provide a rough sketch of the distinct forms that counselling can take, a more contextualized approach is required to identify the ways in which risk and disability are constructed in the counselling setting. A good starting point for such an approach is the work of Rayna Rapp.

In an ethnographic study of prenatal testing and the genetic counselling situation, Rapp has detailed the discursive practices through which genetic counsellors impart information to their women clients (and their partners). According to Rapp, counsellors formulate an opening statement with three goals in mind: first, convey significant information about the risks of birth "anomalies," and about the availability and medico-technical nature of amniocentesis; second, take an individual and family health history; third, communicate with the patient in a manner that allows her questions and concerns to be addressed. Rapp explains that the counsellor's knowledge dominates the interaction intended to achieve the first goal. With respect to the second goal, the patient's knowledge is central; however, the counsellor's medical protocols normatively organize that information. The third goal (namely, appropriate communication) shapes all aspects of the counselling situation.[436]

Rapp notes that genetic counsellors must become science educators to convey information about the risks of birth "anomalies," for their clients are usually unfamiliar or only partly familiar with the information counsellors want to present. The use of metaphors enables counsellors to render intangible objects into tangible ones, supplying images of known objects such as books, pearls, and caterpillars for the karyotype, that indicates chromosomes, paired, and size-ordered. The authority of science is therefore conveyed in what Rapp refers to as "apparently 'user-friendly' descriptions."[437] For, despite the technical nature of the information, counsellors maintain that its importance merits its detailed elaboration. Among the technical information that counsellors convey are the rare reasons why a test might need to be repeated and the potential complications of the test, including the significant (0.5 –1 per cent) risk of a test-induced miscarriage.

The perceived need to impart this technical information to clients can make negotiating language-shifts between audiences of clients tricky business for counsellors. To be sure, it is effective and appropriate to simplify complex information for non-science speakers and to assure that central information is accessible to them. However, this strategy diminishes the base of information upon which the process of decision-making will proceed in these cases. For when non-science speakers receive less scientific information than their more privileged peers, their disadvantaged position is reproduced. Thus, counsellors may be caught between their effectiveness and their thoroughness, that is, between giving too little information or giving too much. While ideally counsellors will assess and solve this problem individually in each case, structural antecedents often forestall personalized solutions. These structural factors reside in the complex intersections of class background and the education resources within which openness to scientific literacy is forged.[438]

## 7.3 Values in Counselling Practice

The question of how to interpret language requirements of a given client raises the issue of counsellor values and objectivity. Sullivan and Heng have remarked that the role of counsellors' values has not received much discussion in the professional literature. Sullivan and Heng point out that most genetic counsellors share the assumption that counselling must be "objective" to foster free, informed, and good decisions, where "objective" signifies avoidance of bias that a counsellor's own values may introduce into a given counselling situation. Sullivan and Heng allow that the ideal of value-free counselling intends to limit the possibility that a counsellor's personal viewpoint will unduly influence the judgment of clients; they argue, however, that the belief that one can simply relay medical "facts" in counselling is naive, because often there is a considerable degree of ambiguity about exactly what the facts are that must be understood.[439] They argue, furthermore, that counsellors should attend to and critically evaluate their personal attitudes and values, rather than strive to ignore or avoid the ways in which their own subjectivity may condition aspects of the counselling situation.

The principle of non-directiveness follows from the belief that if a given counsellor's own moral stance in some way enters into the discussion of the counselling session, then the freedom of the client could be compromised. To promote and respect the dignity and autonomy of clients, therefore, the counsellor's own values are deemed irrelevant and are thus actively suppressed. In an article on prenatal diagnosis, D'Alton and DeCherney assert, for instance that "counselling should be non-directive and should concentrate on the accurate presentation of all the facts and options."[440] For these authors, one of the most important goals of genetic counselling is to ensure that people understand the options available to them. To be sure, D'Alton and DeCherney concede that a variety of personal factors such as previous experience, cultural background, and religious beliefs can influence beliefs that clients hold about the acceptability of diagnostic technologies and the choices that persons make based on these; nevertheless, they refrain from specifying what role, if any, counsellors should play in drawing out the basis of these choices.[441]

Since there is remarkable consensus among genetic counsellors with respect to the desirability of value-free counselling, it would seem to follow that this model of counselling must have merits. Sullivan and Heng suggest four conclusions that seem to support this position.

First, since the goal of a counsellor should be to assist others to make morally responsible decisions, then sufficient knowledge and freedom must be ensured. Thus, the emphasis on factual information is a priority for the counsellor. In addition, the manner in which counsellors present this information is important to promote the autonomy of clients.

Second, the profession recognizes the personal nature of value judgments. The primacy that it places on the value of autonomy derives from the realization that we constitute ourselves by and through our choices. In addition, counsellors recognize that the character of value judgments is quite distinct from the activity of understanding facts and outlining options and consequences. All of these are required in this context if one is to make a free and informed choice. By avoiding the personal dimensions of these issues, counsellors acknowledge the distinctiveness of factual judgments in genetics from value judgments. That counsellors draw this distinction is in addition an effort to restrict counsellors' responsibilities to their area of expertise (namely, factual scientific matters).

Third, the accepted position on value-free counselling indicates sensitivity to the power dynamics of the counselling relationship. Non-directional counselling is recommended to limit the inequalities in experience, knowledge, and presumed authority that are inherent to the counselling context. Insofar as these perceived inequalities are minimized, the possibility that a counsellor will unduly influence the judgments of a given client is thus also limited.

Fourth, attention to the occurrence of bias in one's judgments highlights the need to ensure that one is indifferent to the identities of the specific individuals in any particular case.[442]

While Sullivan and Heng do not dispute these conclusions per se, they nevertheless question whether counselling (or indeed science) is, or ought to be, value-free; in addition, they question the assumption that value-free counselling best promotes client autonomy. They assert that some of the problems that arise in counselling practice with respect to this ideal do so because of the ideal itself, not the counsellors involved in any particular situation. For the very discursive and concrete performance of the counsellor enacts a whole series of values about maternal health, foetal impairment, and even the technology itself.

To be sure, it might be argued that the term "value-free" is meant to refer to the value neutrality of the options that the counsellor presents, not the context within which science or the counsellor works. Perhaps there is a sense in which scientific knowledge is prior to any system of values insofar as it can be used for either good or evil purposes. Sullivan and Heng remark, however, that the notion according to which even scientific knowledge can be considered apart from its application is an idealistic one.[443] For social, political and ideological trends contribute to the direction of research produced in any historical moment and are at the same time influenced by that research and its methodology.[444] In short, medical and scientific endeavours proceed replete with values; thus, the options that counsellors suggest are not really "value-free."

Indeed, Rapp has noted that the counsellor's goal of value-neutrality is a "thorny one." The author remarks that it is hard to make a case for the neutrality of technology explicitly developed to identify and eliminate foetuses with "aberrant" chromosomes. The biomedical and public health interests that have driven the development and routinization of the technology itself evaluate such foetuses as "expendable." While ethicists and counsellors argue that prospective parents have a "right" to know (as well as to not know) the chromosomal status of their foetus and to use that information in whatever way they may wish, the very existence and routinization of the technology does not imply neutrality. To the contrary, that the technology exists and has become routinized assumes that scientific and medical resources should be placed in the service of prenatal diagnosis and potential elimination of foetuses with chromosomal "aberrations."[445] Genetic counsellors themselves, Biesecker and Hamby admit that within the professional discourse, there is an implicit endorsement of abortion as an option for dealing with a variety of genetic chromosomal conditions.[446] In effect, Rapp asserts, counsellors are trained to offer a value-charged technology in a value-neutral manner.

Value neutrality is an even more complex problem when the cross-cultural nature of genetic counselling is taken into account. To some individuals, the doctor (or genetic counsellor) is shirking responsibility by not directing clients toward the "appropriate" choice. Equally, others may regard as unreasonable the assumption that an individual woman alone (and her spouse) must bear the responsibility for decision-making on this matter. In many communities, Rapp explains, elder kin have an important, and often determining role to play in reproductive life. This, she notes, is especially the case with some Asian women, especially recent immigrants from China who speak little or no English. Rapp observed that recent women immigrants from the former Soviet Union and Eastern European countries at times suggested discomfort with the "burden" of individual choice. They were most familiar with a cultural context in which medical experts are expected to interpret the complex facts of a medical situation and recommend the appropriate treatment strategies.[447]

Indeed, it would seem that professional standards boards may not give matters of cultural difference as much weight as they deserve. Rapp reports that all of the genetic counsellors she interviewed indicated there was little discussion in their training of the cultural constraints and resources within which many women and their families must operate. As Rapp points out, however, professional demands make it difficult to include courses on cultural diversity in counsellor training.[448] Put directly, the escalating amount of expertise for which counsellors are responsible is a direct requirement for employment. The lack of attention given to cultural difference within the practice of genetic counselling may, in addition, be reflected in the demographics of the profession itself. Although consciousness of how cultural differences affect the counselling situation is growing, the number of National Society of Genetic Counselors (NSGC) counsellors in the U.S. from African-American, Hispanic, or Asian-American backgrounds remains small at not more than 6 per cent.[449]

In another context, Adrienne Asch and Gail Geller have addressed the issues of value-neutrality and non-directiveness in genetic counselling from the perspective of gender. At present, women predominate in the genetic counselling field.[450] Research conducted on the nature of genetic counselling sessions and client satisfaction with these, Asch and Geller claim, reveals that female geneticists (like other female physicians and health professionals) spend a greater deal of time answering questions and developing a good working relationship with their clients than do males in the same provider role.[451] The data also shows that female counsellors and geneticists tend to be less directive and less opinionated in their discussions than their male colleagues.[452] In addition, female geneticists tend to be more inclined to express concern about the implications of certain decisions for the welfare of a woman's extended family.[453]

Regardless of the gender of the counsellor, or whether the individual is a physician or non-physician, counselling encounters often fall short of transparent, unambiguous dialogue. Like physicians, Asch and Geller write, counsellors can impart subtle biases in a counselling situation through the types of questions they ask, and the information they omit or convey.[454] Indeed, the discourse of genetic counselling is determinedly medico-scientific, which masks and silences some meanings while it creates others.[455] As Asch and Geller point out, women may find a physician's explicit directive counselling no more difficult a thing with which to grapple than a counsellor's covert directiveness in a purportedly non-directive framework. For Asch and Geller, the most serious problems with the communication of genetic information arise from the content of the material itself and the style of its disclosure, especially when the material conveys traditional biases about disability, concentrates merely on biomedical understandings and probabilities, and provides little information about the actual lives of persons with disabilities for whom tests exist.[456]

Tristram H. Englehardt (among others) has written extensively about the value-laden character of medicine. As Englehardt puts it, "medicine medicalizes reality. It creates a world.... Medicine molds the ways in which the world of experience takes shape; it conditions reality for us."[457] Various feminist authors have drawn attention to the ways in which traditional medicine operates in the service of broad political, social, and economic interests and agendas that define, regulate, and control the bodies of women, people of colour, and poor people. For example, Susan Sherwin has argued that medicine constructs an authoritative *medicalized* view of women's experiences that enables it to assume proprietorship over their lives.[458] As Sherwin explains it, the medicalization of functions such as pregnancy and menstruation has placed events that characterize healthy development for women under medical control.[459] For some feminist writers, the "new reproductive technologies" constitute another chapter in a continuing story of the pathologization and medicalization of reproduction – a new and more pernicious means of coercive male appropriation of the female body.[460]

## Risks in Counselling Discourse

Barbara Duden has argued that forms of prenatal diagnosis such as ultrasound "disembody" women and alienate them from their own corporeal experiences of pregnancy. Duden notes that when this sort of argument is raised in a medical context, it usually results in calls for better training and preparation of counsellors so that they may inspire confidence in the benefits of the procedures. Duden wishes to emphasize that the procedures themselves must be questioned from the perspective of their inevitable psychic results on women, not simply from that of their technical efficiency. She asks what do the procedures do to a woman's self-understanding? Duden claims that the "better" the counselling, the more authoritatively and effectively convincing are certain modern ideas, namely that prenatal procedures are good, that pregnancies can be classified, imply risks, and demand supervision. Even a woman who is radically opposed to the termination of pregnancies will submit to an interpretation of herself as in a state that requires "scientific" management. Prenatal programs themselves, Duden asserts, not openness toward abortion, transform a woman's body into a field of operation for technocratic and bureaucratic interventions.[461] Duden points out that sonar screening inflates the "population" deemed at risk in pregnancy. Initially developed for the benefit of women at increased risk, this screening technique is now advocated as a test for every pregnant woman.[462]

Lorna Weir has written that over the last generation, pregnancy has come to be administered under the rubric of risk.[463] Weir explains that prior to the advent of prenatal testing, women who were known to be "at risk" of having a baby with a congenital impairment were given probability estimates of whether the unborn would be affected. At that time, there were few manifest symptoms in the antenatal period recognized as signs of foetal impairment; thus, the chief information of diagnostic use was derived from family histories and estimates of exposure to toxins, carcinogens, etc. Weir argues that the prenatal diagnostic tests of the 1970s and 1980s were distinctively different from prior forms of practice. They were not general statements about the sub-populations to which a given woman was regarded to belong, but rather were predictive at the individual level.[464]

Over the past generation, antenatal care has become increasingly characterized by population-based risk techniques in which the foetal body is the primary surface of emergence – the site for the problematization of risk. Because of this circulation of the problem of risk in antenatal care, the body of the pregnant woman has become the subject of sampling, imaging, and surgical techniques. In short, the category of risk now dominates the process of becoming a mother (Quéniart, 1988). As Weir explains, clinical risk techniques such as prenatal screening and diagnostic technologies implement population-based calculations; furthermore, they form risk groups by applying risk categories to the bodies of certain persons who, in turn, are placed under continuing surveillance for treatment.[465]

In the Canadian context, the attachment of risk to pregnant women is accomplished through three separate processes: standardized risk assessment authorized by each provincial and territorial ministry of health for all women undergoing antenatal care; routine prenatal screening tests for all pregnant women; prenatal diagnostic testing where authorized by indications of "increased risk," either as a result of positive results from screening tests or due to clinical indications.[466] In Ontario, the population of pregnant women is governed under the category of risk through a textually mediated process in accordance with a risk schema designed by the Ontario Medical Association and distributed by the Ministry of Health. In addition, general screening programs such as the one that the province of Manitoba instituted in 1985 govern pregnant women in Canada. It offers a maternal alpha-fetoprotein (MSAFP) blood test to all pregnant women.[467]

It is an axiom of good counselling that a client should be told "her risks" before she decides to take (or refuse) the test. The idea of "risk" as both a quantitative and qualitative concept is central to the information that genetic counsellors seek to convey. Among others things, the language of genetic counselling is intended to enhance awareness of the age-related risk of chromosomal "aberrations." Rapp notes that counsellors seldom speak directly about disability or abortion decisions until and unless some "aberration" is detected. Rather, counsellors describe their goal as "to give reassurance," which in such cases means to return clients to the general population that has a background risk of 2 per cent to 3 per cent of giving birth to a child with an impairment. If older clients forego amniocentesis, they undertake a larger risk because age-related risk is added to background risk. The counsellors that Rapp observed in her study used age charts, graphs or birth ratios to first explain a general risk pattern and then to derive a specific risk ratio based on a pregnant woman's estimated date of delivery. Indeed, Rapp reports that all of the counsellors that she observed employed the language of "background risk," "reassurance," and "added risk." This language, she notes, "foregrounds a statistical, medical, age-related, universal, and wholly individual model of risk."[468] These numbers (and hence the "risk" and "reassurance" discourse that accompanies the technology) are presented as universal, and uninfluenced by other factors such as the general health, nutritional status, or country of origin of the parents.

In addition, this statistical language constructs "iatrogenic anxiety."[469] Although there are approximately 800 genetic conditions that can be diagnosed prenatally, chromosomal "aberrations" are the only screenable ones that systematically increase with age. Older women have long known that there is a greater chance they will give birth to babies with Down Syndrome than will younger women. It is only the current generation of pregnant women that has been told specific risk figures and that associate generic pregnancy anxieties with exact age and statistics. Chromosomal analysis is the only test routinely performed on genetic material in an amniocentesis. Because Down Syndrome accounts for about 50 per cent of the chromosomal differences that this screen detects, Down's looms large in the discourse of genetic counselling.[470]

Advanced age is not the only "risk" that genetic discourse can describe. An increasing number of autosomal recessive conditions (many of which occur at heightened frequencies in specific cultural and national origin groups) are detectable. These include Sickle Cell Anaemia (among people of African descent), Tay-Sachs disease (most prevalent among people of Ashkenazi Jewish background), and Thalassemia (blood diseases most common among Mediterranean and Asian populations).[471] In addition, screening for Cystic Fibrosis (CF), the most common autosomal recessively transmitted disease encountered among those of white European descent, has been piloted. Some available screens are close to 100 per cent accurate (e.g., those for Tay-Sachs or Sickle Cell Anaemia); others, however, probe for conditions caused by a series of functionally related genetic mutations that vary between families and among cultural groups.[472] Screening for the more common CF mutations, for instance, will identify 80 to 90 per cent of "white" carriers and lower percentages of those in other "racial-ethnic" groups.[473] This suggests that carrier and foetal testing nets more statistical probabilities than it does positive or negative diagnoses.

Rapp argues that even when diagnosis is unambiguous, its potential severity requires interpretation. Although some conditions (such as Tay-Sachs) are remarkably consistent in their trajectories, a much more common occurrence is that the positive diagnosis of a given condition cannot predict the "severity" (degree of expressivity) of any particular case, or whether those with the gene will manifest its symptoms (degree of penetrance). Even a 100 per cent accurate diagnosis of Sickle Cell Anaemia does not indicate the extent to which a particular foetus will be affected, nor can a diagnosis of CF or the late-blooming Huntington's disease predict age of onset of symptoms.[474]

The powerful language of risk attached to age, cultural background, and family genetic history requires pregnant women (and possibly their family members) to think about their pregnancies through a sieve of statistics. In doing so, the language of risk muffles other positions and social interpretations from which they might respond to disease and impairment. In short, there is little room for anything that resembles cultural variation in the individually grounded, genetically causative model of risk conveyed by genetic counsellors.[475]

The medical establishment presents genetic counselling as a value-neutral means to elaborate options for action available to enable pregnant women to make decisions regarding testing and its possible outcomes. However, Rapp, Weir, and others suggest that the practice of genetic counselling is a disciplinary mechanism that enlists pregnant women to become *self*-regulating and *self*-governing.[476] They maintain that since prenatal diagnosis is socially constructed in genetic counselling as an inherently good (therefore, "necessary") medical intervention of which a pregnant woman can avail herself, the pressure for her to undergo testing is intensified, and the burden of moral responsibility for the health and "normality" of the foetus that is placed upon her by friends and family, and that she places upon herself, may increase.

Indeed, Anne Waldschmidt contends that genetic counselling is a form of eugenics, practised with the active participation of the individuals concerned once they have been informed of the "facts" and have given their consent. Eugenics has shed its past authoritarian roots and has developed an apparently democratic approach. As Waldschmidt explains, "neo-eugenics" does not need to operate through forms of coercion, pressure, open repression, or control. In other words, the state and society no longer need to urge people to "do their eugenic duty," so to speak. People now "voluntarily" adhere to eugenic lines of reasoning individually, without having been expressly told to do so. For all intents and purposes, neo-eugenics may appear to be more humane than traditional eugenics. Waldschmidt argues, however, that when one considers neo-eugenics more closely, it becomes evident that it is actually a perfected form of the same thing.[477]

Waldschmidt goes on to argue that neo-eugenics functions so well precisely because it is supported and practised "from below," that is, by the woman and the man on the street. It does not need to be enforced from above by the police and the authorities. Not even the human geneticists appear to be acting on their own authority; rather, they seem merely to accord with the wishes of their female clients.[478] The profession of genetic counselling purports to provide pregnant women with information about genetics, genetic testing, risks to their unborn, and options for action that will enable them to make informed choices. However if, as Waldschmidt claims, the practice of genetic counselling is actually a function of eugenic socio-political goals, the choices made within that context will be functional to it as well.

As early as 1969, Robert Sinshiemer had forecast the future for "a new eugenics." According to Sinsheimer, a new eugenics would be implemented on the individual level – a eugenics that operates in the service of "normalcy."[479] Referring to Sinsheimer's argument, Evelyn Keller states that the idea of a "right to normalcy" is "an ideological expansion of molecular biology into both popular culture and medicine."[480] Framing "normalcy" within the language of biomedical "rights" in this way individualizes it. The new eugenics demands that *individuals*, as opposed to the State, take responsibility for arbitrating normality: the new eugenics is choice-driven. The notions that pregnant women have a moral and social responsibility to arbitrate normality, and that future children have "a right to normalcy," rely upon the assumption that "the normal" can be well characterized and selected. Keller claims that this assumption lies at the heart of the new eugenics.[481]

Sinsheimer and Keller's descriptions of the form of a new eugenics reflect a number of the motivational assumptions argued in a recent book on genetics and justice. In *From Chance to Choice: Genetics and Justice*, Allen Buchanan and his co-authors argue that justice allows, and even demands, that "defective" foetuses be genetically corrected and enhanced to enable the persons they would become to "fully participate in the co-operative framework of society." To count as a fully cooperating citizen, one must have an "opportunity range" that is compatible with "normal species-typical functioning." Buchanan and his co-authors contend that disabilities limit opportunities and welfare, even when they are not so severe. They argue that that the lives of persons with disabilities are not worth living, even if those persons do not literally suffer as a result of their disabilities.[482] They argue that foetuses whose disabilities are so severe as to render their future lives unbearable should be eliminated. They also contend that it is a moral wrong for a pregnant woman to carry to term and give birth to a baby with such a genetic makeup (see also Purdy 1995). Non-directiveness about genetic transmission of harmful conditions, they argue, is as indefensible as is non-directiveness about child abuse and neglect. For Buchanan and his co-authors, the norm of non-directiveness in genetic counselling functions as a useful reminder of reasonable disagreement about whether prevention of a particular harmful condition is morally required; however, they maintain that it is indefensible as an inflexible and systematic norm to guide all genetic counselling.

## Risk in a Human Rights Discourse

The argument of *From Chance to Choice* relies upon the assumption that all variation between humans can be attributed to their genetic structure. In this regard, Abby Lippman uses the term *geneticization* to refer to "an ongoing process by which differences between individuals are reduced to their DNA codes, with most disorders, behaviors and physiological variations defined, at least in part, as genetic in origin."[483] Through the process of geneticization, Lippman writes, human biology is incorrectly equated with human genetics, with the implication that the latter acts alone to make us each the organism that we respectively are. In other words, the entire spectrum of human differences will be comprehensible once we have a full understanding of human genetic makeup.[484] This belief is related to an assumption that drives reproductive technology and genetic engineering, namely, that it is desirable to strive to "perfect" human beings. As the authors of *Just Technology* point out, the first of these assumptions diverts attention away from social and environmental factors that contribute to the creation of diversity; in addition, the second assumption violates respect for diversity.[485]

Within the human rights discourse that this publication promotes, the "risks" associated with genetic testing are construed quite differently than is done in standard genetic counselling discursive practices. Geneticization is the reduction of people to their DNA codes and the application of genetic technologies to a range of human conditions and characteristics. This trend may have negative impacts upon persons with disabilities. For instance, this trend and the application of some genetic technologies risk undermining some of the rights that persons with disabilities have recently gained, and reinforcing discrimination against them. In addition, the trend toward geneticization and the application of genetic technologies risk undermining choice and self-determination for people with disabilities.

The authors of *Just Technology* allow that information derived from genetic testing and counselling can, if applied responsibly, be beneficial to prospective parents in their decisions to reproduce and rear children. They point out, however, that it should not be assumed that geneticization will have a positive impact. Geneticization may instead reinforce and perpetuate the traditionally accepted medical model of disability, where disability is framed in the context of individual pathology rather than situated in a particular social context. In addition, renewed acceptance of the medical model and the emerging genetic model of disability risk diverting effort and resources to genetic research and associated endeavours that otherwise would be allocated to the removal of disabling social barriers in the environment. It is these barriers, rather than physical, neurological, or genetic makeup of individuals, that are the sources of disability. In short, human rights discourse holds that the well-being of persons is not dependent on the absence of disability.[486]

According to this view, furthermore, the tendency to geneticize difference poses risks to decision-making with respect to human reproduction. For instance, the availability of procedures of prenatal diagnosis and *in vitro* fertilization risks the commodification of children as consumer objects subject to quality control. Some forms of genetic technology increase the reproductive choices for prospective parents, including parents with disabilities;[487] however, there is more at work here than the choice of whether or not to have children. These procedures make available the option of choosing the qualities of one's children. It must be cautioned that *in vitro* fertilization and embryo diagnosis followed by selective implantation in the womb could conceivably become a common method of selecting embryos with socially desirable qualities, giving rise to a new form of eugenics.[488]

## NOTES

[431] Lupton (1999); see also Beck (1992).

[432] Lupton (1999), p. 14.

[433] See, for instance, Lupton (1994); Douglas (1990).

[434] Browner & Press (1995); Press & Browner (1995); Lippman (1991).

[435] Biesecker & Hamby (2000), pp. 343-346.

[436] Rapp (1999), p. 63.

[437] Ibid., p. 65.

[438] Rapp (1999), p. 66.

[439] Sullivan & Heng (1993), p. 1.

[440] D'Alton & DeCherney (1993), p. 2.

[441] D'Alton & DeCherney (1993), p. 2.

[442] Sullivan & Heng (1993), pp. 3-4.

[443] Sullivan & Heng (1993), p. 4.

[444] International League of Societies for Persons with Mental Handicap (1994).

[445] Rapp (1999), pp. 57-59.

[446] Biesecker & Hamby (2000), p. 346.

[447] Rapp (1999), p. 60.

[448] Ibid., p. 57.

[449] Ibid., p. 60.

[450] Ibid., p. 61.

[451] Asch & Geller (1996), p. 340.

[452] Ibid., p. 341.

[453] Ibid.

[454] Ibid.

[455] Rapp (1999); see also Weir (1998).

[456] Asch & Geller (1996), p. 341.

[457] Englehardt (1986), p. 157.

[458] Sherwin (1992), p. 9.

[459] Sherwin (1992); see also Shildrick (1997); Wendell (1996); Bertin (1995).

[460] Weir (1996), p. 375.

[461] Duden (1993), p. 27.

[462] Duden (1993), p. 27.

[463] Weir (1996), p. 378.

[464] Ibid., p. 379.

[465] Ibid., p. 380.

[466] Ibid.

[467] Ibid.

[468] Rapp (1999), p. 70.

[469] Lippman (1991), p. 31.

[470] Rapp (1999), pp. 70-71.

[471] Ibid., p. 72.

[472] Rapp (1999), pp. 72-73.

[473] Tatsugawa, Fox, et al. (1994).

[474] Rapp (1999), p. 73.

[475] Ibid.

[476] See Rapp (1999); Duden (1991); Lippman (1991).

[477] Waldschmidt (1992), p. 165.

[478] Ibid.

[479] Sinsheimer, quoted in Keller (1992), p. 289.

[480] Keller (1992), p. 291.

[481] Ibid., p. 299.

[482] Buchanan et al. (2000), p. 278.

[483] Lippman (1991), p. 18.

[484] Ibid., pp. 18-19.

[485] International League of Societies for Persons with Mental Handicap (1994), pp. 11-14.

[486] International League of Societies for Persons with Mental Handicap (1994), pp. 10-13.

[487] See, for instance, Kallianes & Rubenfeld (1997).

[488] Ibid., pp. 10-14.

# 8

# SUMMARY OF ISSUES

The paradigm in which genetic counselling is situated is problematic on three counts:

- First, it is premised on a bio-medical definition of health that views genetic difference and disability as "abnormal" and to be eliminated;
- Second, without identifying this inherent bias, and under the authority of medical genetics expertise, it presents itself as value-free, neutral, and non-directive;
- Third, in hiding the inherent eugenic nature of prenatal testing and the complicity of the State and medical professions in working toward this goal, under the guise of giving women "choice," it mandates that women arbitrate normality, compromising their well-being, their human rights, the rights of all people with disabilities, and the rights of children to be free from commodification as consumer objects subject to quality control.

## 8.1 Biomedical Definition of Health

The way in which disability tends to be understood profoundly affects how the information obtained through genetic testing and research will be used. Asch suggests that genetic research is permeated by the medical model of disability – linking every difficulty with the physiological characteristics of the condition and not to any characteristics of the society in which people with the condition live their lives.[489] The attribution of disability to "defects" in genetic structure is a notion underlying much of past and current genetic research and resulting applications. This idea shifts the focus away from the systemic and environmental factors that result in disability. It reinforces the "geneticism" and "geneticization" of health and social policy – where people are seen as "nothing more than the biologically active product of our genes,"[490] and where foetuses and people are labelled and disadvantaged based on genetic information.[491] Lippman, for example, cautions:

> Through this discourse [geneticization], which is beginning seriously to threaten other narratives, clinical and research geneticists and their colleagues are conditioning how we view, name and propose to manage a whole host of disorders and disabilities. Though it is only one conceptual model, "genetics" is increasingly identified as the way to reveal and explain health and disease, normality and abnormality.[492]

Geneticization can have a number of consequences in society in general, and for people with disabilities in particular. These consequences include: the erosion of social cohesion; raising the spectre of the perfect human being, rather than respecting human diversity; neglect of other social causes for disability; the idea of the "healthy il"l; and creation of new barriers to the integration of people with disabilities.[493]

In contrast to the focus on genetic causes of disability, there is now a body of research that critically evaluates traditional research methodology and practice in the biomedical and sociological research on disability and has at its base a theory of disability that takes account of the material constraints in the lives of people with disabilities. In discussing these shifts in the theory and research on disability, Rioux has identified four paradigms in the social and scientific formulation of disability: biomedical, functional, environmental, and human rights approach.[494] There has been a tendency in genetic and other disability-related research to operate within the confines of biomedical and functional paradigms, where the disability is seen as an individual pathology. Authors critiquing the reduction of disability to genetics stress that a biomedical view of disability is not inherently harmful to people with disabilities. It can provide an understanding of a person's genetic differences and possible consequences. It can provide information (e.g. through a diagnosis) at an early stage of a person's life about the particular challenges to be faced in communication, motor, and behavioural development and thus encourages access to early intervention programs and other developmental supports. Such information is vital to a child and to his or her family seeking to nurture as many life chances as possible.

The "harm" in a biomedical perspective comes from using it as the only way of viewing a person. This often leads to the assumption that all, or a significant proportion of the disadvantage experienced by a person with particular conditions arises from genetic or other biomedical conditions. To address the disadvantages that arise from the devalued social and economic status of people with disabilities, a broader perspective for viewing a person is needed – one that sheds light on how the legal system and economic, social, educational, and other environments in which a person lives can determine his/her life chances.

The way that disability is viewed has changed dramatically in the last few decades. Disability is no longer viewed as a medical issue, nor as an issue of a person's capacity to function according to certain standards. Among disability theorists and activists, as well as many policy makers, education professionals, employment experts, and others, disability is seen as a social construct. From this perspective, genetic differences labelled as "abnormal" or "defective" are in fact a natural part of human diversity falling inside the range of "normal" human characteristics. People who have such characteristics often experience barriers to full acceptance and participation in society that may be addressed through supports, services, and inclusive practices and environments.

However, like many people in society, genetic counsellors, medical geneticists, and genetic scientists are not necessarily informed about the social and political issues surrounding disability, nor of the positive contributions and experiences of persons with disabilities and their families. Given the lack of protocols for genetic counselling and practice, and the uninformed public perspective of disability, disability activists and others are concerned that genetic professionals may pressure, or direct parents to avoid the birth of children with disabilities. If the public perceives that disability is not only undesirable, but also preventable, the justification and public support for the delivery of health care and related services to people with disabilities and their families may erode.

## 8.2 Presumption of Value-free Neutrality and Non-directiveness

Norms have been developed to encourage "value-free" genetic counselling.[495] However, the very assumption within a biomedical view that disability represents an abnormality or aberration, rather than a sign of difference and diversity, can contribute to perpetuating a limited understanding of people's potential life chances. This view also limits an understanding of the social, legal, and economic factors that disadvantage people with disabling conditions. Reliance on the principle of "value-neutrality" in genetic counselling may not be an adequate norm in light of the harm that can come to the class of people with disabilities from a perpetuation of devaluing ideas about disability. As well, it has been argued that the principle may be outdated given the critique of assumptions of "value-free" science and "objectivism", and the tendency for reliance on the principle to limit genetic counsellors' critical evaluation of their own values and attitudes.[496] Moreover, the centrality of the principle of "diversity" in the recent *Universal Declaration on the Human Genome and Human Rights* suggests that practices in genetic counselling should take account of this principle in interpreting genetic information and assisting people in making decisions with the information.

## 8.3 Manipulation of the Concept of "Choice"

Genetic testing and screening is considered to be a necessary service in the Canadian health system to avoid the birth of children with certain genetic conditions that have been labelled as "abnormal" and a "burden to families and society." Its importance is now entrenched in law, with doctors who fail to avoid such births being sued for "wrongful" birth and subject to charges of medical negligence and malpractice.

In Canada, women over the age of 35 are provided prenatal screening as part of routine prenatal care. While women are asked to consent to some of the more invasive forms of testing such as amniocentesis and prenatal ultrasounds, other less invasive forms of screening are carried out as a matter of "routine care." In several provinces, routine maternal serum screening (MSS) programs are in place for all women, some of whom are screened without their knowledge or consent. Screening is also recommended to prospective parents if they have a family history of an identifiable genetic condition. Prenatal tests are available for such conditions as Down Syndrome, Fragile X Syndrome, Cystic Fibrosis, and Spina Bifida, among others, and many more are expected to be developed in coming years.

While being told that they are free to decide to carry a "defective" foetus to term, to put the baby in an institution or adoptive home, the pressure to terminate "an affected" foetus is pervasive.

### NOTES

[489] Asch and Geller (1996); Asch (1989).

[490] Somerville (1996).

[491] Rioux (1997); Mykitiuk & Penney (1995); Wolf (1995); Lippman (1991).

[492] Lippman (1991).

[493] ILSMH (1994); Hubbard (1995); Asch (1994).

[494] Rioux (1997).

[495] Knoppers (1995).

[496] Sullivan & Heng (1993).

# 9

# TOWARD A NEW PARADIGM FOR GENETIC COUNSELLING

Currently, no guidelines exist to ensure that genetic counselling takes full account of the potential harms that can come from circulating negative stereotypes of people with disabilities, beyond the "value-neutral" stance. This stance is not adequate for addressing the negative values that have historically attached to people with disabilities and that often appear as neutral in health care practice. For example, triaging access to certain health interventions like heart transplants often systematically restricts access for people with certain types of conditions. The criterion of limited quality of life or limited projected social contribution is yet another example of the negative value given to persons with disabilities.

As part of this research project, a framework for genetic counselling that values all persons was tested in focus groups across the country. In general, there was agreement with the framework, although some caution was expressed around the implications such a framework has on a woman's right to abortion.

As work goes ahead to build consensus around this proposed framework, it will be important to avoid drawing lines between pro-choice and pro-life factions in society. Situating the framework within a human rights discourse that seeks to minimize harm to vulnerable groups in society should help to move the discussion to a different level. Just as we have drawn limits as to what is acceptable practice in new reproductive technologies, including a ban on cloning, so too should limits be set on what constitutes acceptable practice with regards to prenatal genetic testing, counselling, and termination of pregnancies.

Canadians have already drawn limits around foetal selection on the basis of sex. Raising public awareness of the arbitrariness around the identification of conditions that result in disabilities as "abnormal" and the way in which it devalues all people with disabilities, commodifies children and infringes on the human rights of women as they are mandated to arbitrate normalcy should help us to move toward an ethical approach to genetic testing and counselling.

The framework we are proposing helps to move to this new paradigm for genetic counselling. It is built around a:

- Set of shared principles
- Model of ethical practice
- Roles for genetic counsellors.

## Shared Principles

The following principles are based in general on basic human rights that underpin international human rights instruments. Fundamental to this is the notion that actions taken against a foetus labelled as "defective" call into question the worth, dignity, and right to life of all persons with disabilities. Any actions that can cause harm to an entire group in society should be considered inherently unethical.

The principles are:

**1. Dignity**

All persons:

- Have intrinsic dignity
- Have non-instrumental worth because of their humanity, not by virtue of their capacities
- Should be treated as they might be, rather than as they are so that they can become what they might be.

**2. Equality**

All persons:

- Are of equal worth regardless of their health status, intellectual potential, or any other characteristic
- Are entitled to be treated equally. Inequality in treatment should be permitted only if it produces the greatest possible benefits for those least well off.

**3. Diversity**

- Diversity is inherent to humanity
- Every person is genetically unique
- Persons are more than their genes
- Difference is valued. There is no single ideal, perfect or average – what we define as "normal" is subjective or a statistical artefact.

**4. Autonomy**

- Persons achieve their full potential as social beings, and as integral members of families and communities
- Each person is free to refrain from participation in genetic or any other research, screening or testing
- All people have a right to informed consent and decision-making
- Health professionals have a responsibility to respect refusals for treatment; they do not have to satisfy all requests for treatment.

**5. Justice**

- Any interventions to minimize, reduce, or eliminate a specific condition or trait must result in no harm to persons living with that condition or trait
- The duty to protect those who may be vulnerable against oppression is intrinsic to social solidarity
- Socially responsible decisions are those that result in no immediate or long-term adverse impacts on vulnerable members of society
- Parents and society have a duty to value, protect and give unconditional acceptance to all viable children
- Cost-benefit analyses of pre-implantation and prenatal genetic counselling/testing should not include costs of termination as a benefit, but as a wasted opportunity.

## 9.2 Proposed Model of Ethical Practice

The proposed model of ethical practices draws largely on the work of David Turnbull.[497] The model situates people with disabilities and their families at the centre as the most knowledgeable experts about life with a disability. It also puts the onus on the professionals involved to declare their own biases, to impart unbiased knowledge to their clients, and to discuss with them the dangers of eugenic biases.

Thus, an ethical approach to genetic counselling:

- Would require genetic counsellors to disclose their own values and biases, so that persons being counselled are able to choose counsellors whose values match their own. Given that the essence of genetic counselling is values-based decision-making, the counsellor's values would be material to the reasonable patient and therefore, legally should be disclosed to ensure a valid informed consent
- Obtains guidance on the ethics of genetic issues from the knowledge, experience, and lives of people with disability themselves, and from the women and families involved
- Places ethical responsibility on those providing genetic counselling to recognize and understand factors that affect people's decision-making, such as access to and timing of information
- Puts responsibility on professionals to obtain ongoing education about life with disability and to convey that information in a way that values people with disabilities and their families
- Acknowledges that scientific information in the absence of social understanding is incomplete and medically constrained information
- Provides balanced information about personal and familiar experience of life with a given condition and challenges the notion that disability dooms persons and families
- Provides a broad range of information sourced inside and outside of the medical arena to parents concerning: the specific disability; needed services for children with specific disabilities; contacts for a parent support group; and disability rights/independent living organizations.
- Involves professional training that includes:
  - Coursework on the social implications of life with disability for persons and families
  - Coursework on how to recognize and to raise public awareness of eugenic biases throughout the genetic testing and counselling processes
  - Exposure to the principles, laws, disability rights organizations, and peer support groups that constitute the disability rights and independent living movement
  - Where possible, contact with children and adults with disabilities outside clinical settings.

## 9.3 Proposed Roles for Genetic Counsellors

As a critical link between practitioners of medical genetics and pregnant women, genetic counsellors play a significant role in imparting knowledge about genetic conditions. Given the inherent eugenic bias in the field of medical genetics, genetic counsellors should be aware of and challenge these biases as they impart knowledge to their clients. The roles outlined here have been drawn from work by Angus Clarke.[498] Specifically, genetic counsellors should:

- Recognize that an early, precise diagnosis of the condition causing concern enhances knowledge and understanding
- Provide therapeutic benefit for families, even if precise diagnosis has not been made
- Provide information on genetic conditions and their intergenerational transmission to assist in management of the condition and promote well-being
- Provide social and practical support for persons and families with genetic conditions, for those who might develop the condition in the future, and for those whose children have been or may be affected in the future
- Provide information on specific therapies for genetic conditions where they exist
- Reduce "handicap" by working to minimize the stigma associated with the condition with the aim of improving the social status and self-esteem of those affected
- Help to positively shape public attitudes toward people who have been labelled with intellectual and other disabilities
- Help to challenge the eugenic bias inherent in medical genetics.

Given the environment in which medical genetics and genetic counselling takes place, it is clear that guidelines that are at the discretion of professional bodies to implement or not will be insufficient. What is needed is a new paradigm for genetic counselling, in which counselling is regulated outside professions that have a vested interest in advancing eugenic goals. Given the invasiveness of the potential for eugenics throughout genetic testing, research and counselling, it is recommended that a regulatory framework be established that answers directly to parliament.

As well, a process should be put into place to examine where eugenic biases exist within the rapidly emerging field of genetics, of medical genetics and the human genome project itself, so that they can be removed. Awareness and education programs about the inherent dangers of eugenics should be promulgated within university training programs for medical professionals, public policy makers, the legal profession, and the general public.

Ratification of the DPI Solihull Declaration on *The Right to Live and Be Different* by Canada would send a clear signal to people with disabilities that they are valued members of our society. As well, the Canadian government should not ratify the Convention on Human Rights and Biomedicine as some sections are in contravention of the two documents adopted at the 1999 UNESCO Conference on the Sciences.

### *DPI Solihull Declaration*

## THE RIGHT TO LIVE AND BE DIFFERENT

First and foremost we demand:

### *Nothing about us without us*

Up until now most of us have been excluded from debates on bioethical issues. These debates have had prejudiced and negative views of our quality of life. They have denied our right to equality and have therefore denied our human rights.

We demand that we are included in all debates and policy-making regarding bioethical issues.

We must be the people who decide on our quality of life and life experiences.

We also call on our organizations to give support, encouragement and reassurance to those of us who are representing our views on bioethical regulatory bodies.

Particular support must be given to empower the voice of mental health survivors, people with learning difficulties, people who cannot advocate for themselves and disabled children in the debate.

### *Richness in Diversity*

We are full human beings. We believe that a society without disabled people would be a lesser society. Our unique individual and collective experiences are an important contribution to a rich, human society.

We demand an end to the biomedical elimination of diversity, to gene selection based on market forces and to the setting of norms and standards by non-disabled people.

Biotechnological change must not be an excuse for control or manipulation of the human condition or biodiversity.

We recognize that the only way to ensure universal support for and positive recognition of our particular qualities is through stating our right to diversity clearly with good arguments, in open discussion. We must strive for alliances with scientists, the medical professions, ethicists, policy-makers, human rights advocates, the media and the general public.

***ALL Human Beings Are Born Free and Equal in Dignity and Rights***

## THE DISABILITY RIGHTS MOVEMENT SPEAKS OUT[499]

Human rights are the responsibility of the state as well as the individual. Disabled people, our organizations, families and allies must work to ensure that international, regional, and national legal instruments must ensure the implementation of rights throughout all scientific advances and medical practices concerning the human genome, reproduction, assessments of quality of life, therapeutic measures and alleviation of "pain and suffering".

Biotechnology presents particular risks for disabled people. The fundamental rights of disabled people, particularly the right to life, must be protected.

In particular we demand:

An absolute prohibition on compulsory genetic testing and the pressuring of women to eliminate – at any stage in the reproductive process – unborn children who it is considered may become disabled

The provision of full accessible (i.e. jargon-free, easy to read and in alternate media) information from which people can make informed decisions

That European governments do not ratify the Convention on Human Rights and Biomedicine as some sections are in contravention of the two documents adopted at the 1999 UNESCO Conference on the Sciences

That disabled people have assistance to live – not assistance to die

That having a disabled child is not a special legal consideration for abortion

That no demarcation lines are drawn regarding severity or types of impairment. This creates hierarchies and leads to increased discrimination of disabled people generally.

Disabled people must join together in solidarity to ensure our voices in these life-threatening issues.

## STRENGTH IN UNITY

## NOTES

[497] Turnbull (2000), pp. 853-865.

[498] Clarke (1993).

[499] The Council of Canadians with Disabilities (CCD) would like to share for information purposes a statement on biotechnologies prepared by representatives from DPI organizations in 27 countries in Europe, Africa, Australia, and North America.

# 10

# CONCLUSION

With the increasing institutionalization of genetic testing within standard protocols for prenatal care and maternal "risk-management", and with increasing availability of genetic testing, genetic counselling is fast emerging as an integral component of health care systems and services. As the "governance" of pregnancy evolves in an environment of rapidly expanding genetic information, tests, and screening technologies, the policy vacuum that currently surrounds the delivery of genetic testing will begin to be filled. While genetic counselling is usually conceived as a process of communicating information about genetic tests and options for action based on this information, and facilitating a decision-making process, its implications are far-reaching. Concerns have been raised that expansion of genetic testing brings with it a risk of a "new eugenics."

> Our propensity to judge each other based on attributed characteristics will increase our potential for intolerance and the appearance of an Animal Farm philosophy in which some are more equal than others. Different characteristics will be targeted, depending on the cultural, political, philosophical, economical and spiritual background of any given society.[500]

Procedures and guidelines for balancing what can be conflicting mandates have not been fully addressed in genetic counselling. These mandates could be characterized, in a preliminary way, to include: respecting value-neutrality and other established norms of genetic counselling (confidentiality, consent, etc.); promoting the principle of diversity in understanding disability; predicting what a disabling condition will mean in a person's and family's life; promoting autonomous decision-making; limiting the harms that can come to the class of persons with disabilities through state-funded and regulated health care decision-making processes, and interventions based on devaluing assumptions about disability.

## NOTES

[500] Wolbring (d/u).

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